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**Case Report**

# A rare case of limb girdle muscular dystrophy with predominant scapulo-humeral involvement: diagnostic challenges and multidisciplinary management in a middle-aged male

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## ABSTRACT

Limb-girdle muscular dystrophy (LGMD) is a genetically and clinically heterogeneous group of neuromuscular disorders characterized by progressive proximal muscle weakness and varying patterns of scapular winging. Accurate subtype identification is often hindered by diagnostic complexities and limited access to advanced diagnostic tools, particularly in resource-constrained settings. We present the case of a 38-year-old male with a 10-year history of progressive upper limb weakness, scapular winging, and persistent fatigue. Electromyography (EMG) revealed a myopathic pattern, supporting the clinical suspicion of LGMD. Muscle biopsy and genetic testing were not performed due to patient unwillingness and financial limitations. A multidisciplinary management approach comprising low-dose corticosteroids, physiotherapy, and nutritional support led to symptomatic improvement and enhanced functional capabilities. This case underscores the diagnostic challenges posed by the absence of advanced genetic and histopathological investigations and highlights the importance of accessible, patient-centered care. Scapular winging and proximal weakness were pivotal diagnostic features in this case, reflecting the hallmark manifestations of LGMD. The observed improvement with physiotherapy and corticosteroids demonstrates the value of a tailored management strategy in mitigating the impact of this progressive condition. This case emphasizes the need for broader access to diagnostic innovations and advanced therapeutic options to optimize care for patients with rare neuromuscular disorders.

**Keywords:** LGMD, Proximal muscle weakness, Scapular winging, Electromyography, Resource-limited settings, Multidisciplinary management, Neuromuscular disorders, Patient-centered care

## INTRODUCTION

Muscular dystrophies (MDs) represent a heterogeneous group of inherited disorders characterized by progressive muscle weakness and degeneration. These conditions exhibit remarkable clinical diversity, ranging from early-onset severe forms to milder, late-onset variants, with considerable variation in genetic etiology and pathophysiological mechanisms. Muscular dystrophies are traditionally classified based on genetic mutations affecting structural or functional proteins within muscle fibers, resulting in progressive degeneration and regenerative failure of skeletal muscle tissue. Among the subtypes, limb-girdle muscular dystrophy (LGMD) stands

out as a particularly diverse and complex group of disorders, primarily affecting the proximal limb-girdle musculature and encompassing over 30 genetically distinct subtypes.[1] LGMD is characterized by a heterogeneous clinical spectrum, including progressive weakness, scapular winging, and functional impairments that significantly impact the quality of life. The condition is further classified into autosomal dominant (LGMD1) and autosomal recessive (LGMD2) subtypes, each associated with specific genetic mutations. Advances in molecular diagnostics, particularly next-generation sequencing (NGS), have expanded our understanding of LGMD's genetic basis, revealing both common and rare mutations responsible for its various subtypes.[2] Despite these advancements, the clinical-genetic overlap among LGMD subtypes and with other neuromuscular disorders, such as spinal muscular atrophy (SMA) and facioscapulohumeral dystrophy (FSHD), poses significant diagnostic challenges. This overlap complicates subtype identification, delays diagnosis, and increases the overall cost and complexity of patient management.[3,4] A key diagnostic feature in LGMD, scapular winging, results from weakness or paralysis of the scapulothoracic stabilizers, such as the serratus anterior or trapezius muscles. This clinical sign serves as a critical diagnostic clue, highlighting functional impairments like difficulty in arm elevation and abduction, which are hallmark symptoms in many neuromuscular disorders. Scapular winging not only reflects the progression of muscular involvement but also underscores the debilitating impact of the disease on daily activities.[5,6] Imaging modalities, including magnetic resonance imaging (MRI), have emerged as valuable tools in assessing scapular winging and monitoring muscle pathology, complementing the clinical evaluation and electrophysiological studies.[7,8] The diagnostic process for LGMD exemplifies the importance of a multidisciplinary approach. The integration of clinical examination, imaging, electromyography (EMG), and genetic testing is crucial for accurate diagnosis and differentiation from other neuromuscular disorders. Recent advances in NGS and whole-exome sequencing (WES) have significantly enhanced the diagnostic yield for LGMD by enabling simultaneous analysis of multiple candidate genes. These technologies have proven particularly effective in resolving diagnostic dilemmas associated with overlapping phenotypes and have facilitated the identification of novel pathogenic variants.[9,10] In one study, WES improved the diagnostic success rate for undiagnosed LGMD cases by 45%, highlighting the transformative potential of genetic sequencing in clinical practice.[11] Despite these advancements, several challenges remain. Variants of uncertain significance (VUS) and the lack of well-established genotype-phenotype correlations hinder the interpretation of genetic data and complicate clinical decision-making. Furthermore, disparities in access to advanced diagnostics and genetic counselling pose additional barriers, particularly in resource-limited settings.[12] Multicenter studies have underscored the need for standardized diagnostic algorithms that incorporate clinical, histological, and molecular data to improve diagnostic accuracy and patient outcomes.[13] The use of quantitative imaging techniques, such as dixon-based MRI, has shown promise in tracking disease progression and evaluating therapeutic efficacy in LGMD subtypes like LGMD2I, offering a non-invasive alternative to traditional biopsy methods.[14] In the context of diagnostic challenges and the clinical diversity of LGMD, detailed case reports play a pivotal role in advancing our understanding of rare and atypical presentations. They provide invaluable insights into the natural history, diagnostic features, and management strategies for this group of disorders.

The present report describes a unique case of LGMD with predominant scapulo-humeral involvement, emphasizing the diagnostic utility of clinical evaluation, imaging, and genetic testing in distinguishing LGMD from phenotypically similar conditions. By contributing to the growing body of literature, this case underscores the importance of an integrative approach in addressing the diagnostic complexities of LGMD and highlights opportunities for improving patient care through advances in molecular diagnostics and imaging technologies.

## CASE REPORT

A 38-year-old male day laborer presented with a 10-year history of progressive weakness in both upper limbs, accompanied by persistent generalized fatigue. The patient first noticed difficulty in raising his arms above his head, carrying heavy objects, and holding his arms in an outstretched position. Over time, these difficulties worsened, significantly impairing his ability to perform occupational tasks and daily activities. Despite this, fine motor functions such as writing or grasping objects remained preserved. He later observed visible muscle wasting in the upper limbs, with underlying bones becoming prominent.

The patient reported no weakness in the lower limbs, difficulties with walking or balance, or diurnal variation in symptoms. He denied any associated facial muscle weakness, drooping eyelids, swallowing problems, or changes in vision, hearing, or voice. His bowel and bladder habits were normal. There was no history of chest pain, palpitations, shortness of breath, or peripheral edema.

### *Past medical history and personal history*

The patient was normotensive, non-diabetic, and non-asthmatic, with no significant past illnesses or surgeries. He was a non-smoker and non-alcoholic.

### *Family and developmental history*

There was no history of similar illnesses in the family, consanguinity, or sudden deaths. The patient reported normal childhood development with no delays in achieving milestones.

***Examination findings***

The patient appeared cachectic and malnourished, with below-average build. Vital signs were within normal limits. General examination revealed no pallor, jaundice, cyanosis, or lymphadenopathy.

Neurological examination showed significant muscle wasting in the proximal upper limbs, with evidence of scapular winging confirmed by a positive wall push-up test. Muscle power grading (Medical Research Council scale) revealed - upper limbs: proximal muscle groups 2/5, distal groups 5/5, and lower limbs: proximal muscle groups 4/5, distal groups 5/5. Muscle tone was normal in all extremities, with no fasciculations, grip myotonia, or percussion myotonia. Reflexes were preserved, including biceps, triceps, supinator, knee, and ankle jerks. Plantar response was equivocal. Sensory examination, including fine and crude touch, vibration, joint position, and proprioception, was intact bilaterally.

Systemic examinations of the cardiovascular, respiratory, gastrointestinal, and musculoskeletal systems were unremarkable, except for scapular winging and proximal upper limb muscle wasting. No other deformities, contractures, or joint abnormalities were noted.

***Laboratory and initial investigations***

Routine laboratory tests showed - elevated serum creatine phosphokinase (CPK): 350 U/l (normal: 24–195 U/l), normal serum aldolase: 3.58 U/l (reference: ≤7.6 U/l), and normal complete blood count, serum creatinine, and random blood glucose. Chest X-ray, electrocardiogram (ECG), and echocardiogram were normal, showing no signs of cardiac involvement.

***Provisional diagnosis***

Based on clinical and laboratory findings, the provisional diagnosis was LGMD, predominantly scapulo-humeral variety.

***Differential diagnoses***

It included: late-onset spinal muscular atrophy (SMA), and facioscapulohumeral muscular dystrophy (FSHD).

***Management plan***

*Diagnostic approach*

Given the constraints of the patient's circumstances, a tailored diagnostic approach was implemented.

*Electromyography*

Conducted to evaluate neuromuscular involvement, revealing a myopathic pattern that aligned with the clinical suspicion of LGMD and excluded neurogenic disorders such as spinal muscular atrophy (SMA) or peripheral neuropathies.

*Magnetic resonance imaging*

Suggested to assess the cervical spine and screen for structural or compressive abnormalities, though specific results were not detailed.

*Nerve conduction velocity studies*

Recommended to evaluate peripheral nerve integrity and assist in differential diagnosis; results remain unspecified.

*Muscle biopsy*

Not performed due to the patient’s unwillingness, limiting the ability to obtain histopathological confirmation.

*Genetic testing*

Not carried out due to financial constraints, reflecting challenges in achieving definitive subtype identification in resource-limited settings.

***Symptomatic management***

*Low-dose corticosteroids*

Administered to address symptoms, leading to some functional improvement observed during follow-up. Although corticosteroids are not standard for all LGMD subtypes, they were used here as a feasible option for symptomatic relief.

*Physiotherapy*

A structured regimen focusing on strengthening exercises was implemented to preserve muscle function, enhance mobility, and prevent joint contractures. Regular physiotherapy sessions contributed to noticeable improvements in the patient’s daily functional capabilities.

*Nutritional support*

Interventions were provided to address cachexia and malnutrition, aiming to improve the patient’s overall health and resilience against disease progression.

*Patient counseling*

Comprehensive counseling sessions were conducted to educate the patient about the progressive nature of LGMD, set realistic expectations, and discuss long-term care and treatment options.

***Follow-up and monitoring***

Regular follow-up visits were planned with specific goals.

*Monitoring disease progression*

To assess changes in muscle strength and functional capabilities over time.

*Evaluating response to treatment*

To track improvements or adjustments needed in corticosteroid therapy and physiotherapy.

*Addressing emerging symptoms*

To identify and manage any new complications or limitations arising from disease progression.

***Future considerations***

While genetic testing and advanced diagnostics were unavailable in this case, the patient was counseled about potential future opportunities, including clinical trial participation and emerging therapies like gene therapy. These discussions highlighted the critical need for expanded access to diagnostic and therapeutic innovations in resource-constrained settings.

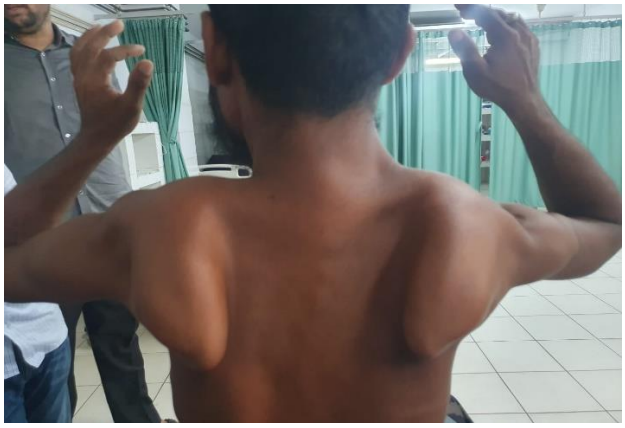

**Figure 1: Scapular winging in a patient with suspected limb-girdle muscular dystrophy - posterior view during active arm elevation.**

**Figure 2: Positive wall push-up test highlighting scapular winging in a patient with suspected limb-girdle muscular dystrophy.**

## DISCUSSION

This case highlights the diagnostic and management challenges of LGMD, particularly the scapulo-humeral subtype, underscoring its clinical heterogeneity, progressive nature, and significant impact on quality of life. The patient presented with hallmark features of LGMD, including progressive proximal muscle weakness, scapular winging, and elevated serum CPK levels, aligning with findings from similar cases reported in the literature. Scapular winging, a defining characteristic in our patient, is frequently observed in LGMD and reflects dysfunction of the scapulothoracic stabilizers, leading to significant functional impairments such as difficulty with arm elevation and lifting. The clinical relevance of scapular winging is well-documented, emphasizing its role as both a diagnostic clue and a determinant of daily activity limitations.[5,15]

The differential diagnosis in this case posed a considerable challenge due to overlapping clinical features with other neuromuscular disorders such as late-onset SMA and FSHD. These disorders share phenotypic similarities, including proximal muscle weakness and scapular winging, complicating the initial clinical evaluation. Advanced diagnostic tools, including EMG, genetic testing, and muscle biopsy, were pivotal in narrowing the differential diagnosis. Literature supports the indispensability of genetic testing for definitive diagnosis in LGMD, as it enables the identification of specific gene mutations and helps differentiate LGMD subtypes from phenotypically similar conditions.[16-18] Studies have further emphasized the value of muscle biopsies in cases where genetic results are inconclusive, highlighting their role in elucidating the underlying myopathic processes.[19]

The progressive nature of LGMD significantly impacts patients' quality of life, particularly due to mobility restrictions, chronic fatigue, and social limitations. Studies evaluating the quality of life in LGMD patients reveal that fatigue and mobility issues are primary determinants of reduced independence and overall life satisfaction.[20] This aligns with our patient's reported difficulties in performing occupational tasks and basic daily activities, such as raising his arms and carrying objects. Management strategies in LGMD focus on preserving functional abilities and mitigating complications. Physiotherapy, emphasizing strength and endurance training, has shown benefits in maintaining muscle function and delaying disease progression.[21] Our patient was prescribed a multidisciplinary rehabilitation program, which is consistent with recommendations for managing LGMD, emphasizing physiotherapy, nutritional support, and symptomatic treatment.[22]

Emerging therapeutic modalities, such as gene therapy and cell transplantation, hold promise for altering the course of LGMD. Recent advances in clinical trials utilizing gene-editing tools, such as CRISPR-Cas9, and viral vectors for gene replacement therapy underscore the potential for

targeted interventions to modify disease progression.[23,24] However, access to such therapies remains limited, and their applicability is constrained by subtype-specific considerations. Our patient's participation in a comprehensive care plan, including genetic counseling and exploration of clinical trials, exemplifies the critical role of early and personalized interventions in improving outcomes and quality of life.

This case also underscores the importance of patient-centered approaches in LGMD management. Qualitative studies have highlighted the value of addressing patient-reported outcomes, such as mobility, emotional well-being, and social functioning, in developing effective care strategies.[25] For our patient, counseling was a key component of management, helping set realistic expectations and enhance psychological resilience in the face of a progressively debilitating condition.

In conclusion, this case exemplifies the complexities of diagnosing and managing LGMD, particularly its scapulo-humeral variant. It underscores the importance of a systematic, multidisciplinary diagnostic approach combining clinical evaluation, advanced molecular techniques, and functional assessments. While current management strategies focus on symptom alleviation and functional preservation, emerging therapies hold promise for modifying disease trajectories. Comprehensive, patient-centered care remains central to optimizing outcomes and addressing the multifaceted challenges posed by LGMD.

### *Limitations*

The primary limitation of this study lies in the absence of genetic testing and muscle biopsy, which are essential for the definitive diagnosis and precise subtyping of LGMD. Financial constraints and patient unwillingness posed significant barriers to performing these investigations, limiting the scope of diagnostic confirmation. Additionally, advanced imaging, such as muscle MRI, was not conducted to further delineate the extent of muscle involvement and identify specific patterns associated with LGMD subtypes. The management approach was constrained by resource limitations, precluding access to emerging therapeutic options such as gene therapy or participation in clinical trials. These limitations reflect the challenges often encountered in resource-limited settings and underscore the importance of improving accessibility to advanced diagnostic and therapeutic tools for neuromuscular disorders.

## CONCLUSION

This case highlights the complexities involved in diagnosing and managing LGMD, particularly in resource-constrained settings. The diagnosis was primarily based on clinical presentation and electromyographic findings, with the absence of definitive genetic or histopathological confirmation. Despite these constraints, a multidisciplinary approach combining low-dose corticosteroids, physiotherapy, nutritional support, and patient counseling led to symptomatic improvement and enhanced functional capabilities. The case underscores the value of tailored, patient-centered care and the need for broader access to advanced diagnostics and therapeutic options. Future efforts should focus on addressing these gaps to optimize outcomes and improve the quality of life for patients with rare neuromuscular disorders.

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