

Research Article

# Clinico-pathological diagnosis of Facioscapulohumeral Dystrophy in a 22-year-old Male

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## Abstract

**Background:** Facioscapulohumeral dystrophy (FSHD) is a rare hereditary disease with a prevalence of 2.03–6.8 per 100,000 individuals. FSHD is the third most common type of muscular dystrophy after the Duchene muscular dystrophy and myotonic dystrophy. To the best of our knowledge, the current case report is the first to report probable FSHD case mainly diagnosed using clinico-pathological evidence from sub-Saharan Africa (SSA).

**Case Report:** A 22-year-old right-handed male college student presented with progressive proximal muscular weakness associated with wasting. The weakness started from the bilateral facial muscles and progressively involved proximal upper and lower limbs muscles associated with scapular winging, waddling gait, and bilateral foot drops. His bulbar, sensory, autonomic, and cognitive systems were spared. Muscles EMG showed myopathic patterns and normal serum CK. Muscle biopsy from affected muscles showed variation in fiber size with groups of angular fibers, preserved fibers, and hypertrophic fibers with marked fibrosis and adipose tissue replacement with no apparent inflammation and necrosis which is consistent with pathological features of muscular dystrophy. Considering the clinical semiology, physical findings, EMG findings, and pathological findings diagnosis of FSHD of scapulo-peroneal variant was made. The patient was managed with analgesics, nutritional advice, and ankle prosthesis for foot drops. Currently, the patient is in a similar condition with modest improvement in his musculoskeletal pain complaints.

**Conclusion:** This case highlights the fact that a careful clinical evaluation with thorough utilization of diagnostic investigations available at our disposal may support the diagnosis of FSHD in resource-limited areas where the necessary genetic tests were not available.

**Keywords:** facioscapulohumeral muscular dystrophy, dystrophy, clinico-pathology, sub-Saharan Africa

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## 1. Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is a rare hereditary disease with a prevalence of 2.03–6.8 per 100,000 individuals [1]. FSHD is the third most common type of muscular dystrophy after the Duchene muscular dystrophy and myotonic dystrophy [1, 2]. The pathology of FSHD is a result of an intricate interaction of genetics, involving the protein product of the *DUX4* gene and the chromosomal location and number of repeats of the D4Z4 microsatellite. The result of this interaction is the inappropriate expression of the *DUX4* protein product; however, it is sporadic in 30% [1–3]. FSHD is classified into two categories: FSHD 1 and FSHD 2 [1]. The vast majority (95%) of patients meeting clinical criteria for FSHD will have FSHD1, while the remaining 5% constitute for FSHD 2 [1].

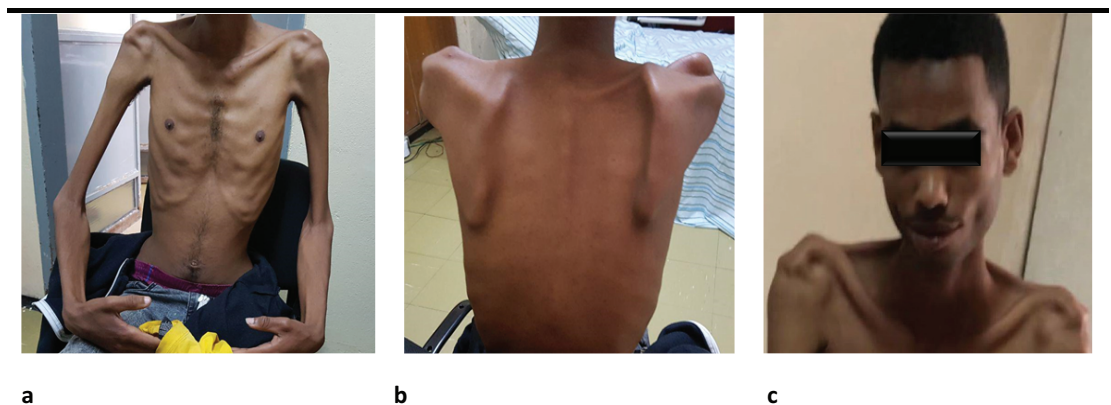
The disease often onsets in the second decade of life; however, the onset during infancy and adulthood have also been reported [4]. The muscular weakness initially involves proximal muscles such as facial, shoulder girdle, and arm muscles and subsequently progresses downward to involve lower limb and pelvic girdle muscles; however, FSHD relatively spares extra ocular, pharyngeal, lingual, respiratory, and the myocardial muscles compared to other muscular dystrophies [1, 3, 5–8]. Diagnoses of FSHD depends on the fulfillment of clinical criteria, genetic study, electromyographic (EMG), and histopathological evidence [1, 2, 4–7, 9, 10]. To date, there is no proven disease-modifying therapy for FSHD; however, supportive treatments such as nutritional advise, spine alignment surgery, and ankle prosthesis could be beneficial to patients with FSHD [1, 4, 10, 11]. To the best of our knowledge, this is the first case report on the diagnosis of FSHD based on clinico–pathological pieces of evidence from sub-Saharan Africa (SSA).

## 2. Case Report

A 22-year-old male presented with progressive weakness and wasting (atrophy) of muscles of the bilateral face, shoulder, arm, pelvic girdles, and distal limb over a four-year period. In addition, he had associated atrophy of proximal muscles of shoulder and pelvic girdles. Progressively, he developed difficulty in combing and washing his hair, standing from a sitting position, and waddling gait. Furthermore, he reported severe back and joint pain which worsened when he walked and relieved with rest due to his Hyperlordosis. Otherwise, no mentation changes, cognitive impairment, bladder or bowel complaint, and no bulbar dysfunction were reported. The patient had no family history of similar illness, no previous history of tuberculosis treatment. He is a third-year

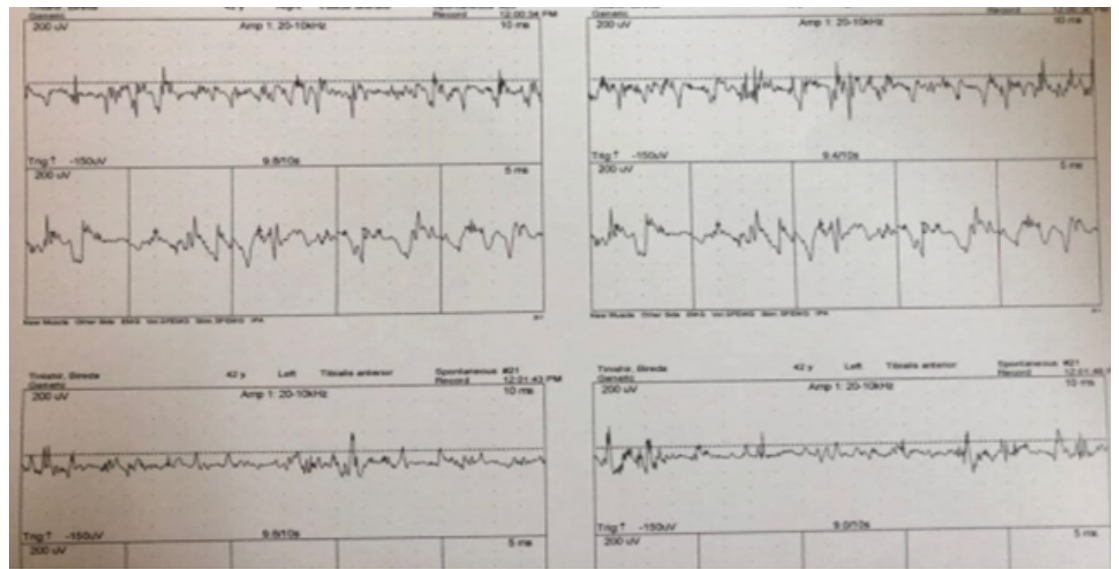
university student but discontinued his education because of progressive extremities weakness and gait abnormality. The patient lives with his mother and one younger brother, both of whom are healthy. His father died when he was young, cause unknown.

General appearance was emaciated. Vital signs were normal. Atrophied shoulder muscles, bilateral pectorals muscles, intercostal muscles, proximal arm and pelvic muscles with relative preserved muscle bulk of bilateral forearm and bilateral scapular winging were noted (Figures 1a and 1b). The patient had prominent lordosis and positive Gower sign. Neurological examination showed bilateral facial diplegia (Figure 1c), atrophied muscles with a power of 3/4 in proximal muscles and 4/5 in distal muscles in both upper limbs. Bilateral foot drop was noted. Reflexes, tone, sensory examination, and plantar reflexes were normal. Routine investigations were normal. Serology tests for HIV and HBSAg were negative. Creatinine kinase was normal. Echocardiography (ECHO), electrocardiography (ECG), and nerve conduction test (NCS) were normal. Electromyography (EMG) showed myopathic patterns without features of active denervation potentials (Figure 2). Muscle biopsy from affected muscles showed variation in fiber size with groups of angular fibers, preserved fibers, and hypertrophic fibers with marked fibrosis and adipose tissue replacement with no apparent inflammation and necrosis which is consistent with the pathological features of muscular dystrophy (Figures 3a and 3b).

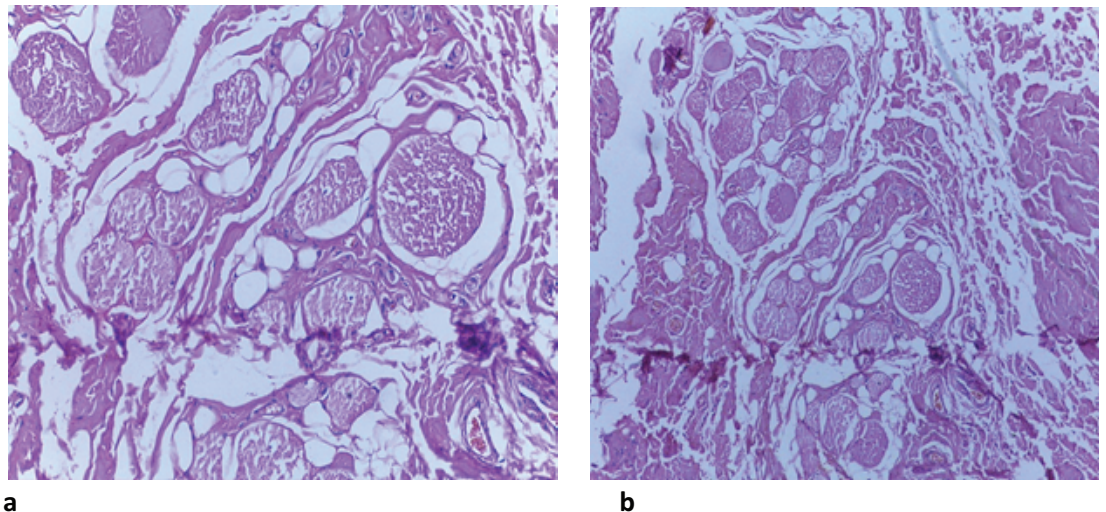


**Figure 1:** (a) Image showing significant muscular atrophy of intercostal, pectoralis, arm, and shoulder girdle muscles. (b) Image showing bilateral scapular winging while the patient extends his upper limbs. (c) Image showing slim elongated face with modest facial diplegia.

Considering the clinical semiology and the physical, EMG, and pathological findings, the diagnosis of FSHD of scapuloperoneal variant was made. A definitive diagnosis of FSHD requires genetic study confirming mutations in a *SMCHD1* gene. However, genetic studies were not available in Ethiopia and if at all available, are not affordable by the patients. Therefore, we solely based the diagnosis of FSHD in our patient on the typical clinical presentation, EMG findings, and histopathological evidence. The patient and his



**Figure 2:** Electromyography (EMG) image showing small amplitude and duration Muscle Action Unit Potential (MUAP) suggesting myopathic patterns.



**Figure 3:** E&M muscle biopsy from affected muscles showed variation in fiber size with groups of angular fibers, preserved fibers, and hypertrophic fibers with marked fibrosis and adipose tissue replacement with no apparent inflammation and necrosis which is consistent with pathological features muscular dystrophy.

family were advised on the available treatment options and prognosis of the disease. He was started with NSAIDs for his back pain and joint pain. In addition, nutritional advice was given in order to improve his muscle bulk. Ankle prosthesis was prescribed for his bilateral foot drop. Currently, the patient is in a similar condition with modest improvement in his musculoskeletal pain complaints.

### 3. Comments

We present a young male who presented with progressive proximal muscular weakness and wasting. The weakness started from the bilateral facial muscles and progressively involved proximal upper and lower limbs muscles associated with scapular winging, waddling gait, and bilateral foot drops. His bulbar, sensory, autonomic, and cognitive systems were spared. Muscles EMG showed myopathic patterns and normal serum CK, and histopathological examination from affected muscles revealed muscular dystrophy. His overall evaluation suggested FSHD. Since genetic tests were not done, FSHD classification was not settled. The patient was managed symptomatically. FSHD commonly occurs in the second or third decade of life [5]. Our patient is a 22-year-old male, which, in line with the typical age of FSHD symptoms onset. We did conduct a genetic study of our patient because of the unavailability of genetic tests. Despite the absence of a positive family FSHD history in our patient, the fact that his father died of unknown cause could remotely support the underlying genetic etiology. However, nearly 30% of FSHD patients could be sporadic [12].

Accurate diagnosis of FSHD requires presences of hallmark clinical semiology such as proximal onset muscle weakness and wasting (facial diplegia) and progressive involvement of shoulder, pelvic, and distal lower limbs muscles, sparing ocular, bulbar, sphincter, respiratory, and cardiac muscles [1, 3, 6, 7, 13, 14]. Likewise, our patient's clinical features started with bilateral facial weakness which resulted in difficulty of closing his eyes, whistling, and blowing chicks. Furthermore, progressive weakness and wasting of shoulder, intercostal, and arm muscles resulted in scapular winging. In addition, pelvic girdle muscles and distal legs muscles weakness resulted in waddling gait and foot drops, respectively. Myopathic pattern, EMG findings, and normal or elevated serum CK often support the diagnosis of FSHD [7]. Our patient's EMG findings were suggestive of myopathic pattern. However, the serum CK of our patient was normal, which could be an expected possibility in patients with FSHD. Muscle biopsy is important in order to support the diagnosis of FSHD in resource-limited areas like us. Histopathological analysis from affected muscles could indicate angular muscle fibers atrophy and scanty inflammatory cells, which will help us differentiate FSHD from other inflammatory myopathies [1]. Therefore, the combination of clinico-pathological evidence and EMG findings could be highly valuable in diagnosing muscular dystrophies such as FSHD in resource-limited regions such as SSA. In the absence of supportive genetic evidence for FSHD, it is important to rule out other dystrophies with similar clinical features such as Emery–Dreifuss muscular dystrophy (EDMD), a slowly progressive X-linked muscular

dystrophy with a late onset and a slightly reduced average lifespan [15]. The classic overall triad of EDMD consists of early contractures, progressive muscle weakness and atrophy, and cardiac abnormalities. The absence of muscular contracture and normal cardiac function in our case makes the diagnosis of EDMD less likely.

Currently, the treatment options of FSHD are limited to symptomatic managements such as relieving musculoskeletal pain, spine-alignment surgery, ankle prosthesis, and nutritional advice [1, 5, 6]. Thus far, our patient was treated with analgesics for his musculoskeletal pain, nutritional advice to increase his lean body mass, and ankle prosthesis for his foot drops. Spine alignment surgery for Hyperlordosis was not done, as the service was not available in Ethiopia. This case highlights the fact that a careful clinical evaluation with thorough utilization of diagnostic investigations available at our disposal may support the diagnosis of FSHD in resource-limited areas where the necessary genetic tests are not available.

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## Statement of Ethics

The author's institutions does not require ethical approval letter for publication of a single case. The patient has given written informed consent for publication of the case. The copy of the written consent will be available from the corresponding author upon reasonable request from the journal editor.

## Competing interests

The authors have no conflicts of interest to declare.

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None

## Authors' contributions

All authors were involved in the conception, writing and critical review of the study. All authors were involved in patient's evaluation, investigation, treatment, and follow-ups. The final version of the article has been approved by all authors for publication.

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