Pakistan Journal of Neurological Sciences (PJNS)

Volume 19 | Issue 1 Article 4

3-2024

Atypical Facio-Scapulo-Humeral Dystrophy with Facial Sparing: A Case Report

Umer Younas
Combined Military Hospital, Hyderabad, Sindh, Pakistan

Asma Aamir
Army Medical College, Rawalpindi, Pakistan

Farooq Azam Rathore
Armed Forces Institute of Rehabilitation Medicine (AFIRM), Rawalpindi, Pakistan

Follow this and additional works at: https://ecommons.aku.edu/pjns

 Part of the Neurology Commons

Recommended Citation

Younas, Umer; Aamir, Asma; and Rathore, Farooq Azam (2024) "Atypical Facio-Scapulo-Humeral Dystrophy with Facial Sparing: A Case Report," *Pakistan Journal of Neurological Sciences (PJNS)*: Vol. 19: Iss. 1, Article 4.
Available at: https://ecommons.aku.edu/pjns/vol19/iss1/4

https://doi.org/10.56310/pjns.v19i01.296

ATYPICAL FACIO-SCAPULO-HUMERAL DYSTROPHY WITH FACIAL SPARING: A CASE REPORT

Umer Younas[1], Asma Aamir[2], Farooq Azam Rathore[3]
[1].Combined Military Hospital, Hyderabad, Sindh, Pakistan
[2].Army Medical College, Rawalpindi, Pakistan
[3].Armed Forces Institute of Rehabilitation Medicine (AFIRM), Rawalpindi, Pakistan

Corresponding Author: Umer Younas *Combined Military Hospital, Hyderabad, Sindh, Pakistan* ***Email:*** *umeryounassp@gmail.com*

Date of submission: *October 11, 2023* ***Date of revision:*** *March 2, 2024* ***Date of acceptance:*** *March 22, 2024*

ABSTRACT

Facio-Scapulo-Humeral Dystrophy (FSHD) is an autosomal dominant and third most common inherited muscular dystrophy. Typical phenotype of FSHD initially affects the muscles of the face and upper arm. Atypical FSHD refers to genetically confirmed FSHD with phenotypic variations. Scapulo-Humeral Dystrophy (SHD) is the most common atypical form of FSDH characterized by no facial muscle weakness on clinical examination, while other signs remain consistent with FSHD. We report a 31-year-old male who presented with foot drop and had an unexpected presentation as SHD. Provisional diagnosis of SHD was based on careful medical history, physical examination findings, modest rise in muscle enzymes, and needle electromyography findings of myopathy. Muscle biopsy suggested dystrophic changes which supported the diagnosis of FSHD with facial sparing. Rehabilitation plan included patient and family counseling/education, lifestyle modifications, energy conservation strategies, right ankle foot orthosis, and therapeutic exercises with regular follow-up for monitoring of the disease.

KEYWORDS: Muscle disorders, Peripheral neuromuscular disorders, Atypical Myopathy, Scapulohumeral dystrophy

INTRODUCTION

Muscular dystrophies refer to a group of inherited, progressive myopathic disorders resulting from defect in genes needed for normal muscle cell structure and function.[1,2] Facio-Scapulo-Humeral Dystrophy (FSHD) is a form of muscular dystrophy with autosomal dominant inheritance. It is the third most common inherited muscular dystrophic myopathy after dystrophinopathies and myotonic dystrophy.[3] It was first described by Landouzy and Dejerine in 1884 and has an estimated incidence of more than 1:10000.[4]

FSHD characteristically involves face and upper arm musculature. Atypical FSHD refers to genetically confirmed FSHD with phenotypic variations.[3,4] (Figure 1) Facial-sparing Scapulo-Humeral Dystrophy (SHD) is the most common atypical form of FSDH with no facial muscle weakness on clinical examination; while other signs remain consistent with FSHD.[3-6] Although prevalence of FSHD in Pakistan is unknown, however, a few case reports of typical FSHD have been reported at national level.[7-9] So far, no case of SHD (FSHD with facial sparing) has been reported at national level. We report a young male who presented with non-traumatic foot drop, as an unusual presentation of SHD.

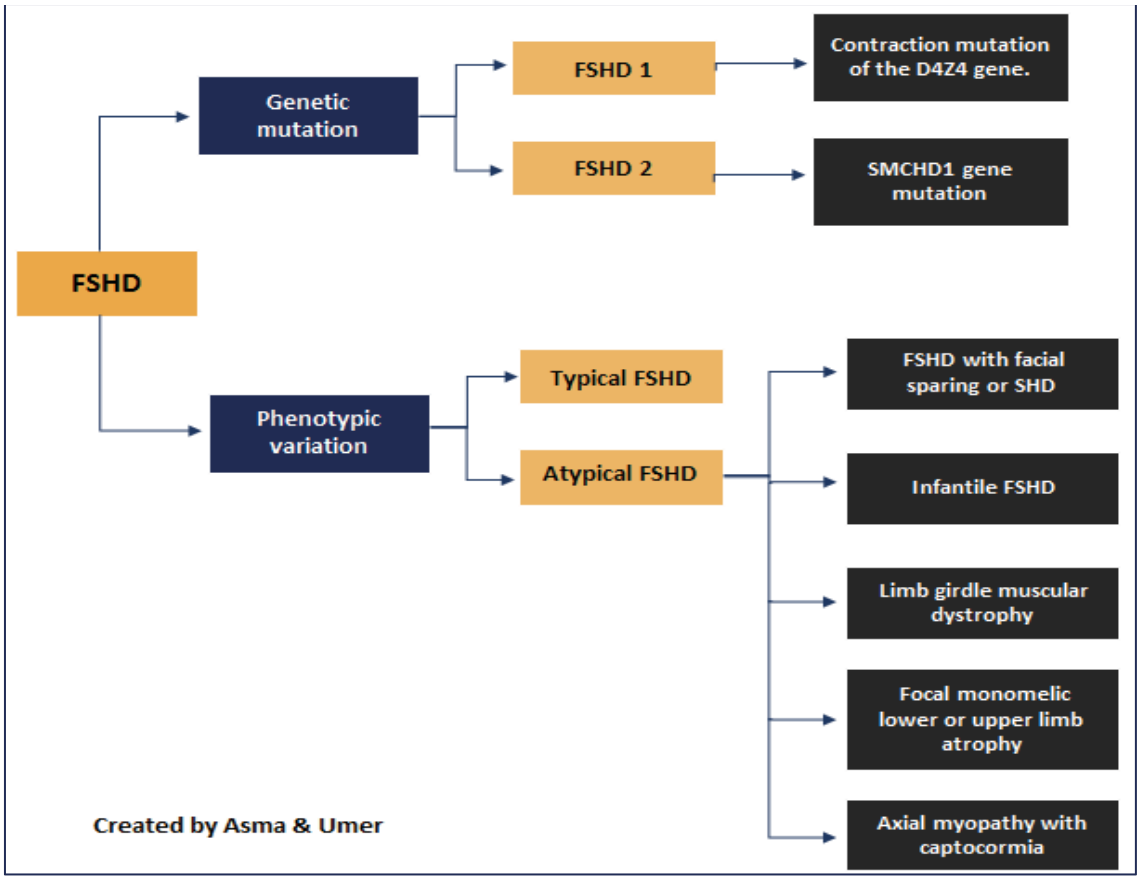

Figure 1: Types of FSHD based on genetic mutation and phenotypic variation

CASE PRESENTATION

A 31-year-old male right-hand dominant, truck driver presented to Combined Military Hospital, Hyderabad with right foot drop for 4 weeks. He developed painless, slowly progressive weakness of the right leg over the last five months. This resulted in a road traffic accident after which he stopped driving. Past medical history revealed that he was well until 17 years of age and worked as a laborer when he first experienced difficulty in lifting blocks with his right arm. Weakness was progressive, painless, and associated with wasting of right arm muscles. Inability to perform overhead activities with his right arm resulted in a change of profession to truck driving. Four years ago, he started experiencing similar progressive weakness and wasting in his left arm resulting in difficulty in activities of daily living including combing hair, bathing, and dressing upper body. The patient experienced difficulty in rising from the floor which gradually worsened over the last 9 months. However, he continued driving until he met a road traffic accident and finally decided to get a medical consult. There was no history of developmental delay, back pain, bladder or bowel dysfunction, fever, chronic steroid use, difficulty drinking from a straw, dysphagia, dysphonia, hearing issues, or family history of similar medical conditions.

On examination, there was symmetric wasting of biceps, triceps, and pectoral muscles with relative deltoid sparing, horizontal anterior axillary folds, scapular winging, prominent trapezii, and relative wasting of the right calf (Figure 2). Muscle bulk of the forearm and intrinsic hand muscles was preserved. There was no facial muscle weakness or ptosis and the patient was able to whistle, smile, and pout the lips without difficulty. He easily buried his eyelashes when asked to close eyes tightly (Figure 3). Active range of motion (ROM) at shoulder was restricted to 90 degrees of forward flexion and abduction. Passive ROM of all joints was full and pain-free. On manual muscle testing, powers were 3/5 in bilateral biceps, triceps, and deltoid, 5/5 in bilateral wrist extensors and flexors and intrinsic hand muscles, 3/5 in bilateral gluteus medius and maximus, 4/5 in bilateral hip flexors and knee extensors,

2/5 in right ankle dorsiflexors and foot evertors, 4/5 in right plantar flexors, and 5/5 in all muscles of the left leg and foot. Beevor's sign was positive suggesting weakness of lower abdominal muscles. Gower's and Trendelenburg sign were also positive suggesting weakness of hip girdle muscles. The patient moved with a combination of high steppage and waddling gait with a hyperlordotic back (compensating for right foot drop and weak glutei). Sensations, deep tendon reflexes, and fine hand movements were intact with downgoing plantars. Systematic examination was unremarkable.

Figure 2: Wasting of scapular and humeral muscles with relative sparing of deltoids.

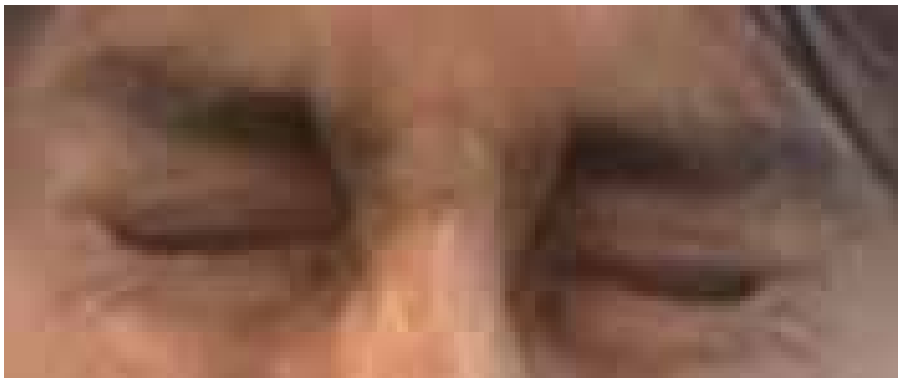

Figure 3: Intact power of Orbicularis Oculi – feature of facial sparing

Serum creatine kinase 492 U/L (25-170 U/L) was raised. Sensory and motor nerve conduction studies were normal. Needle electromyography showed small, short polyphasic motor unit action potentials with early recruitment and full interference pattern in biceps, triceps, deltoid, trapezii, glutei, and right tibialis anterior and gastrocnemius. Muscle biopsy of left triceps brachii revealed dystrophic features. Genetic testing, spirometry, and 2D-Echo were declined by the patient due to cost issues.

Based on clinical history, physical examination, and available investigations, a diagnosis of SHD, i.e., FSHD with facial sparing was made. The patient was counseled about the nature of the disease and advised to avoid heavy physical exertion, consider lifestyle modifications, energy conservation strategies, right ankle foot orthosis, and therapeutic exercises. At the 3-month follow-up, he had a better insight into the disease and a slight functional improvement.

DISCUSSION

To the best of author's knowledge and online literature search, this is the first documented case of SHD from Pakistan. Patient had foot drop as an unusual presentation. There are two subclasses of FSHD based on genetic mutations (Figure 1). FSHD 1 is the most common form and comprises 95% of the cases. FSHD 2 is less common and comprises 5% of cases. [2, 3]

FSHD is phenotypically classified into typical / classic and atypical forms (Figure 1). The classic form is characterized by slowly progressive, asymmetrical weakness of facial (striking clinical feature), scapular, upper arm (humeral) muscles which progresses to lower leg (dorsiflexors), abdominal muscles and later may involve pelvic girdle. Patients become symptomatic in second decade and by 20 years, clinical findings are seen in up to 90% of patients.[3,4] FSHD with facial sparing or SHD is the most common atypical form of FSHD, characterized by very small 4q35 deletions and a slow progression.[5, 6, 10] Patients with SHD are able to forcefully close eyes, burry eyelashes, pucker lips, puff out cheeks which suggests intact function of orbicularis oris and oculi muscles.[6,7] However, other clinical features like scapular winging, wasting of the upper arm (humeral) muscles with relative sparing of deltoid (Popeye's arms appearance), ankle dorsiflexors and pelvic girdle involvement, are consistent with typical FSHD. [4, 6, 10] Our patient had clinical features typical of SHD.

Extra-muscular manifestations may include pain, fatigue, hearing loss (especially of high frequency sounds), vascular retinopathy, cardiac involvement (cardiac myopathy and dysrhythmia), kyphoscoliosis (resulting in respiratory insufficiency). [3, 5, 10]

DNA testing is the gold standard for diagnosis of FSHD. [4, 5, 10] Other diagnostic tests include serum creatine kinase levels, serum aldolase levels, muscle biopsy, nerve conduction studies/ electromyography (NCS/EMG) and muscle magnetic resonance imaging. [3-10] In familial cases, examination of the family members is also considered an important diagnostic criterion.[4] Our patient had slightly raised muscle enzymes. NCS/EMG was suggestive of myopathy and muscle biopsy revealed dystrophic changes. Genetic testing was not done due to cost issues.

Currently, there is no definitive cure for this disease.[5, 7, 8, 10] However, timely diagnosis and early start of multidisciplinary rehabilitation interventions may help in slowing disease progression, preserving functional abilities and improving quality of life. Depending upon the disability of the patient, a multidisciplinary rehabilitation team can include rehabilitation medicine physician (team leader), neurologist, physiotherapist, occupational therapist, psychologist, nutritionist and social worker. Pulmonologist, cardiologist, and orthopaedic surgeon consultation is also required. Rehabilitation approaches include patient and family education, lifestyle and environmental modifications, preservation of muscle strength and function, monitoring for secondary complications and disease progression, energy conservation techniques, assistive devices (like ankle foot orthosis) and gait aids [4-7, 10] Surgical intervention requires prior careful evaluation and realistic goal setting. Surgical options include scapulopexy and scapulodesis which improves scapular fixation to the thorax.[7,8]

CONCLUSION

Due to phenotypical variations at time of presentation and rare occurrence, SHD has a high likelihood of being misdiagnosed. In absence of genetic testing, careful documentation of medical history taking, meticulous neurological examination and relevant investigations (muscle enzymes, NCS/ EMG, muscle biopsy) remain most important tools for timely diagnosis. Early rehabilitation interventions help in preserving / improving function and quality of life. Regular monitoring and follow ups can help in minimizing preventable secondary complications and slowing the progression of the disease.

REFERENCES

1. Myopathy [Internet]. www.ninds.nih.gov. NIH National Institute of Neurological Disorders and Stroke; 2022 [cited 2022 Dec 10]. Available from: https://www.ninds.nih.gov/health-information/disorders/myopathy
2. Cifu DX. Braddom's Physical Medicine and Rehabilitation. Sixth Ed. Philadelphia: Elsevier; 2021. Chapter 42, Myopathic Disorders; p. 885-894.
3. Hassan A, Jones LK, Milone M, Kumar N. Focal and other unusual presentations of f acioscapulohumeral muscular dystrophy. Muscle Nerve. 2012 Sep;46(3):421-5.
4. Krasnianski M, Eger K, Neudecker S, Jakubiczka S, Zierz S. Atypical phenotypes in patients with facioscapulohumeral muscular dystrophy 4q35 deletion. Arch Neurol. 2003;60(10):1421-5
5. He JJ, Lin XD, Lin F, Xu GR, Xu LQ, Hu W, et al. Clinical and genetic features of patients with facial-sparing facioscapulohumeral muscular dystrophy. Eur J Neurol. 2018;25(2):356-64.
6. Pastorello E, Cao M, Trevisan CP. Atypical onset in a series of 122 cases with FacioScapuloHumeral Muscular Dystrophy. Clin Neurol Neurosurg. 2012;114(3):230-4.
7. Gill ZA, Rathore FA, Waheed A, Ahmed N. Facioscapulohumeral muscular dystropy: case report. Pak Armed Forces Med J. 2012;Jun 30(2):135
8. Ikram M, Ayaz SB, Gill ZA, Rathore FA. Facioscapulohumeral muscular dystrophy; A report of. Rawal Med J. 2014;39(1):96-9
9. Aftab S, Hamid MH, Sultan T. A rare case of myasthenia gravis with coexisting muscular dystrophy. Pak J Neurol Sci. 2014;9(2):22-3.
10. Hamel J, Tawil R. Facioscapulohumeral muscular dystrophy: update on pathogenesis and future treatments. Neurotherapeutics. 2018 Oct;15(4):863-71.

Acknowledgement:
We acknowledge the patient for his consent to share his story and case report with the scientific community.

Disclaimer: It was presented as a poster in 1st International Student Research Conference held at Aga Khan University, Karachi from 24-26 Mar 2023.

Conflict of interest: Author declares no conflict of interest.
Funding disclosure: Nil

Authors' contribution:
Umer Younas; concept, case management, manuscript writing
Asma Aamir; case management, manuscript writing
Farooq Azam Rathore; case management, manuscript revision
All the authors have approved the final version of the article and agree to be accountable for all aspects of the work.

This is an Open Access article distributed under the terms of the Creative Commons Attribution-Non Commercial 2.0 Generic License.