International Journal of Contemporary Pediatrics
Sinha R et al. Int J Contemp Pediatr. 2025 Oct;12(10):1718-1720
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20252973

Case Report

Interesting signs in facioscapulohumeral muscular dystrophy in a child

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Received: 26 June 2025
Revised: 01 August 2025
Accepted: 11 September 2025

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ABSTRACT

Facioscapulohumeral muscular dystrophy is the third most common muscular dystrophy in the world after Duchenne muscular dystrophy and myotonic dystrophy. The age of onset of FSHD varies from infancy to late adulthood. The prevalence ranges from 1:8000 to 20,000, with sixty percent of the disease population in Asian population. We report interesting signs in a genetically proven case of Facioscapulohumeral Muscular Dystrophy in a 9 years old child who had presented with facial muscle weakness, shoulder girdle weakness followed by hip girdle weakness. The clinician should be aware of this rare genetic condition especially when there is asymmetric weakness of facial and shoulder girdle muscles.

Keywords: Beevors, Dystrophy, Fascio scapular, Humeral, Polyhill

INTRODUCTION

Facioscapulohumeral muscular dystrophy is the third most common muscular dystrophy in the world after Duchenne muscular dystrophy and myotonic dystrophy. The age of onset of FSHD varies from infancy to late adulthood. The prevalence ranges from 1:8000 to 20,000, with sixty percent of the disease population in Asian population.[1] There are 2 genetically distinct types of FSHD, FSHD1 and FSHD2.[2] It is an autosomal dominant condition with genetic anticipation in several generations of a family. The genetic defect is a loss of a critical number of repetitive elements (D4Z4) in the 4q sub telomeric region.[3] We report interesting signs in a genetically proven case of Facioscapulohumeral Muscular Dystrophy in a 9 years old child who had presented with facial muscle weakness, shoulder girdle weakness followed by hip girdle weakness.

CASE REPORT

A 9 years old boy presented with facial, shoulder girdle weakness followed by hip girdle weakness of 2 years duration. He was not able to frown, blow balloons, close his eyes and was expressionless when laughing and crying. The clinical course was slowly progressive. There was no related family history. The clinical examination revealed wasting of temporalis, trapezius, deltoid, biceps, triceps muscles. The proximal muscle strength of upper limbs and lower limbs were grade 3 and 4 respectively. There was asymmetrical scapular winging (right >left) with positive polyhill sign (Figure 1) and positive Beevor's sign (Figure 2).

There was no calf hypertrophy and grip myotonia. The eye and hearing evaluation were normal. His intelligence quotient was 95 which was normal. His creatine phosphokinase was 1200 IU/l (Normal 120-180 IU/l). His optical genome mapping for D4Z4 gene revealed repeat count <2 (Normal repeat range>11) confirming the diagnosis of Facioscapulohumeral Muscular Dystrophy (FSHD) type 1. Parents were counselled and supportive physiotherapy started with light aerobic exercise and stretching regimen to prevent disuse atrophy. Weaknesses are very slow progressive and child is able to swim, ride bicycles and attend school with minimal disability.

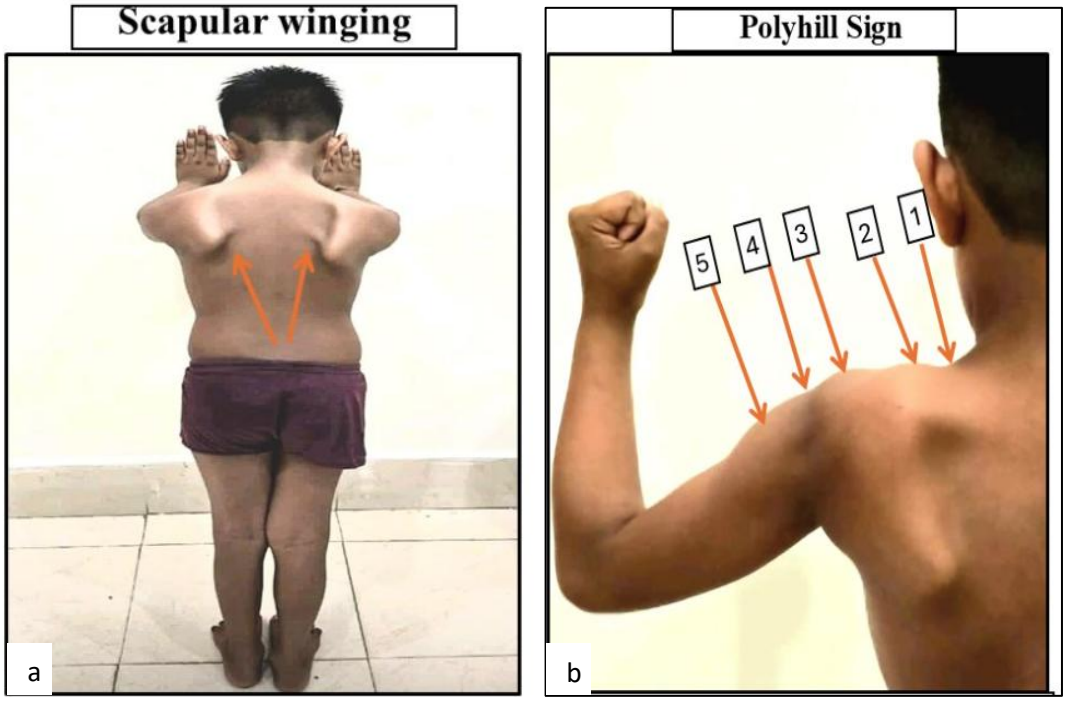

Figure 1: (a) Scapular winging right more than left when patient pushes against the wall with extended arms. (b) Poly-hill sign (from the back). Arrow 1: wasting of trapezius, arrow 2: superior angle of the scapula, arrow 3: displaced of the acromioclavicular joint, arrow 4: atrophied proximal deltoid and arrow 5: normal bulk of distal deltoid.

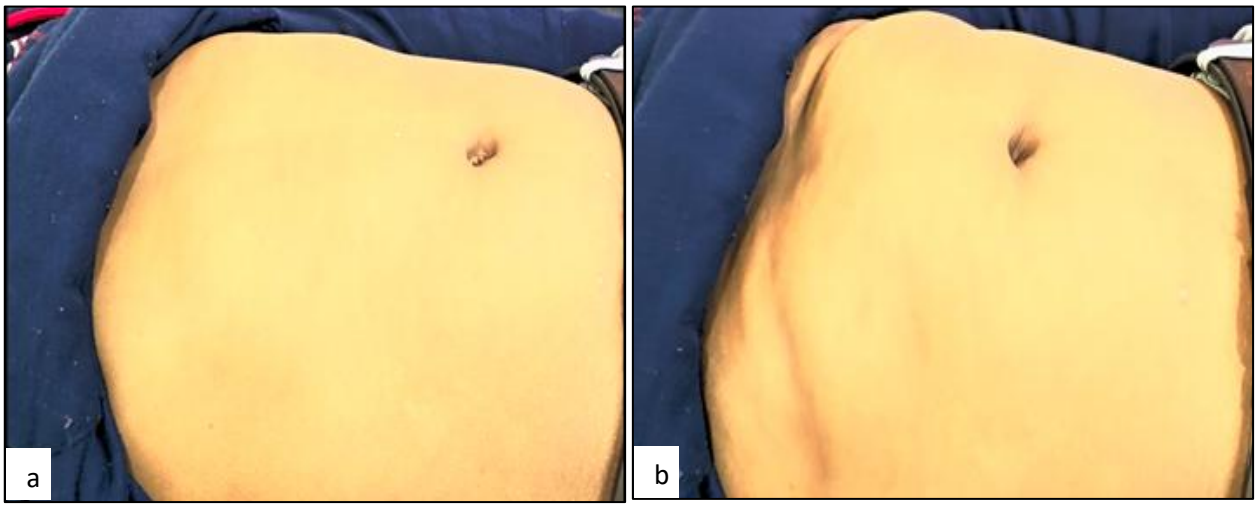

Figure 2: (a) Normal position of umbilicus when patient is lying supine. (b) Abnormal upward movement of the umbilicus on attempting to raise the head from a supine position suggestive of weakness of inferior portion of the rectus abdominal muscle.

DISCUSSION

Facioscapulohumeral muscular dystrophy is the third most common muscular dystrophy in the world after Duchenne muscular dystrophy and myotonic dystrophy.

The age of onset of FSHD varies from infancy to late adulthood. The genetic defect is a loss of a critical number of repetitive elements (D4Z4) in the 4q sub telomeric region.[3] The D4Z4 repeat sequence contains the DUX4 gene, which is abnormally expressed in skeletal muscle cells of FSHD patients.[4] The abnormal expression of DUX4 leads to dysregulation of molecular pathways that are involved in muscle differentiation, oxidative stress responses, immune responses and protein turnover.[5,6]

Early involvement of facial and scapular muscles and subsequent involvement of proximal muscles (including the biceps and triceps) and final involvement of the

pelvic girdle muscles are typical manifestations of FSHD. Gradually most of the affected patients will have multiple clinical signs including eye closure weakness, reduced facial expression, transverse smile, inability to suck from a straw, periscapular muscle atrophy, high riding scapula, horizontal clavicles, Beevor's sign, increased lumbar lordosis as well as hip girdle and foot extensor weakness.[7] Atypical features include relative facial sparing, calf hypertrophy and predominantly lower extremities weakness. There is presently no effective pharmacological treatment for FSHD.

CONCLUSION

The clinician should be aware of this rare genetic condition especially when there is asymmetric weakness of facial and shoulder girdle muscles.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Sinha R, Pathak A. Interesting signs in facioscapulohumeral muscular dystrophy in a child. Int J Contemp Pediatr 2025;12:1718-20.