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A Rare Case of Faciosacpulo Humeral Muscular Dsytrophy

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Abstract

Facioscapulo humeral muscular dystrophy (FSHD) is a rare type of myopathy and differs clinically and genetically from Duchene muscular dystrophy . we present a case of (FSHD) in a 30 years old male. Contrary to the typical autosomal dominant inheritance pattern, this case appears sporadic in nature. FSHD usually manifests in adolescence, and while the condition predominantly affects the face, its slow progression means symptoms may also be observable in children.

Keywords: Facioscapulo Humeral Muscular Dystrophy, Myopathy, Autosomal Dominant Disorder

Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is a rare form of muscular dystrophy with an estimated prevalence from 1/8000 to 1/20000, inherited in autosomal dominant manner (1). The vast majority of FSHD patients exhibit deletions in the subtelomeric repeat array known as D4Z4, located on chromosome 4q35. While normal individuals posses between 11 to 150 repeats of this array, FSHD patients typically have fewer than 11(2,3). Interestingly, these deletions do not appear to disrupt any identifiable gene. Rather, they cause the telomere to be positioned closer to the centromere, indirectly affecting the expression of neighboring genes. The exact mechanism underlying the disease remain unknown, but it is speculated to involve aberrant chromatin interactions at the nuclear envelope. Somatic mosacism maybe prevalent, particularly in de novo cases. Herein, we present a case of FSHD in a 30 yrs old male with no prior family history of the condition. He started to experience difficulty to maintain erect posture since 6 years which progressed gradually over time till today.

Case report

A 30 yrs old gentleman presented with history of weakness in arms and face with difficulty in maintaining

erect posture since 6 years. There was no history of fever, pains, paraesthesia or any trauma in the past with negative family history.

On examination he was well built, normotensive with normal general physical examinations. CNS examination showed normal mental function and fundus examination was also normal. He has bilateral facial weakness with bilateral sensory neural hearing loss, he has exaggerated lord tic attitude while standing and walking with difficulty in getting up from floor .there is marked shoulder muscle wasting and winging of scapula more on left side and prominence of scapula was more evident when he tried to push against the wall with elbows extended and hands at shoulder level. (Fig1&2)

Fig.1 Facial weakness with difficulty in pouting and smile.

His investigations revealed normal hemogram with normal liver and kidney function, his total CPK was raised 199 units per lits. , EMG was done which showed myopathic pattern (fig 3). Muscle biopsy showed features of myopathy with paraspinal muscle atrophy.

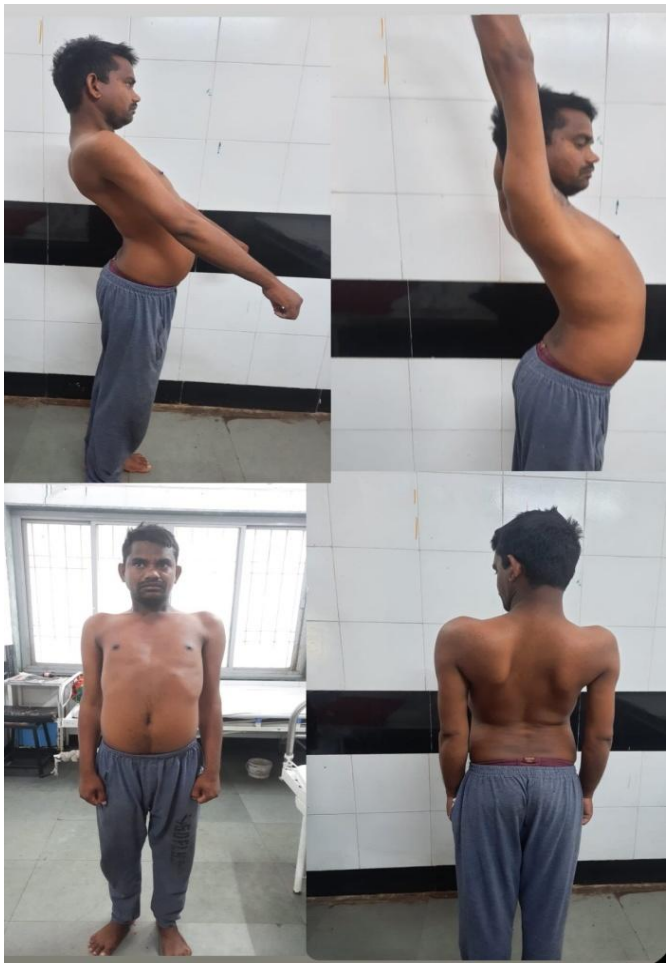

Fig.2: Depicting exaggerated lumbar lordosis and winging of scapula

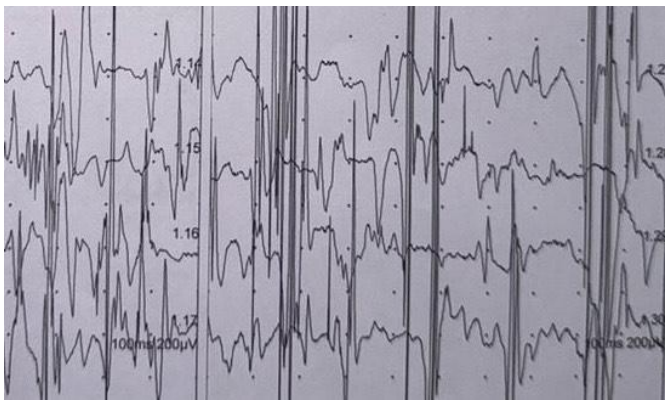

Fig.3: Depicting EMG showing myopathic pattern

Discussion

Facioscapulohumeral Muscular Dystrophy (FSHD) is a relatively uncommon disorder characterized by autosomal dominant inheritance. This means that inheriting just one copy of the defective gene from one

parent is adequate to cause the condition. Typically, FSHD manifests during adolescence, though symptoms may be apparent earlier in childhood. Initially, muscle weakness may be uneven, and individuals with milder forms of the disease may not even realize they are affected. However, FSHD can progress over time, with approximately 15-20% of patients eventually requiring wheelchair assistance, and a small fraction may necessitate ventilatory support as the condition advances.

In the case described, a 30 Yrs old male presented with weakness in the face and arms. Notably, there was no family history of the condition, indicating that this particular case was sporadic rather than inherited.

FSHD may also present with additional features such as mild sensorineural hearing loss and vascular retinopathy, although these manifestations are not consistent among all affected individuals, and their relationship to the genetic abnormality underlying FSHD remains unclear.

Overall, this passage provides an in-depth overview of FSHD, covering its clinical presentation, associated features, and the variability in its progression and presentation among individuals.

Conclusion

Facioscapulohumeral Muscular Dystrophy (FSHD) is a rare muscle disorder that should be considered in children displaying muscle weakness. It's crucial not to mistake it for oculopharyngeal muscular dystrophy, which typically manifests much later in life, usually between the ages of 40 and 60.

Currently, there is no specific treatment available for FSHD. However, scapular stabilization procedures have demonstrated efficacy in improving scapular winging, a common symptom associated with the condition.

Given the absence of definitive treatment options, the primary focus should be on providing comprehensive counseling to both the parents and patients regarding the nature of the disease, its anticipated progression, and available strategies for managing symptoms to enhance overall quality of life. Physiotherapy remains the mainstay strengthening therapy for limb and facial muscle weakness.

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