CASE REPORT

Facioscapulo Humeral Muscular Dystrophy

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Abstract

Facioscapulo humeral muscular dystrophy (FSHD) is a rare type of myopathy and differs clinically and genetically from Duchenne muscular dystrophy. It is an autosomal dominant disorder and as the name designates it has the characteristic distribution of the weakness. The face is almost always affected. Progression of the disease is slow and symptoms begin in adolescence but signs may be evident in children. We present a case of facioscapulo humeral muscular dystrophy in a 14 year old girl & the case was sporadic in nature.

Key Words

Facioscapulo Humeral Muscular Dystrophy, Myopathy, Autosomal Dominant Disorder

Introduction

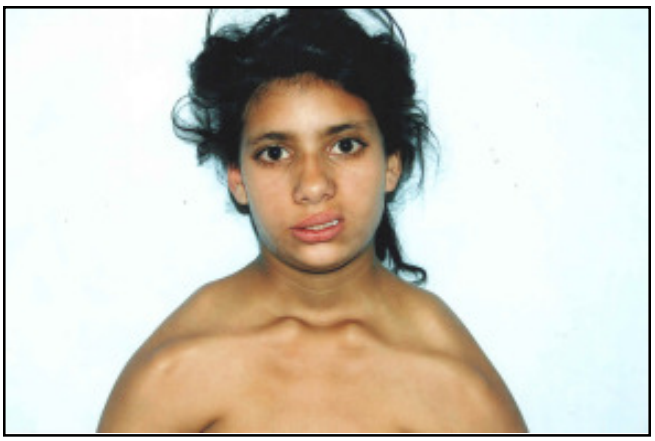
FSHD is a rare type of muscular dystrophy with autosomal dominant inheritance (1). Almost all FSHD patients have deletions of the subtelomeric repeat array, termed D424, on chromosome 4q35. Normal individuals have 11 to 150 repeats; FSHD patients have fewer than 11 (2, 3). The deletions donot seem to interrupt any identifiable gene. Instead, they make the telomere closer to the centromere to seem to act indirectly, increasing the expression of neighboring genes. The pathogenesis of the disease is unknown but may result from inappropriate chromatin interactions at the nuclear envelope. Somatic mosaicism may be common in denovo cases. We report a case of FSHD in a 14 year Old girl who has negative family history.

Case Report

A 14 year old girl presented with history of weakness of arms & face since one year duration. There was no history of fever, pains, paraesthesias or any trauma in the past with negative family history.

On examination she was well built, normtensive with normal general physical examinations. C.N.S examination showed normal mental functions and fundus examination was also normal. She has bilateral facial weakness with no other cranial nerve involvement. She was unable to close her eyes, smile or whistle. There was marked wasting of shoulder girdle muscles with winging of the scapulae and prominence of the scapulae was more evident when she tried to push against the wall with elbows extended and hands at shoulder level. Her shoulder gridle had a characteristic appearance when viewed from the front, the clavicles seems to sag and the tips of the scapulae project above the scapulo clavicular fossa. There was also weakness of abdominal muscles (*Fig 1 & 2*) .

Fig. 1 Characteristic Appearance of Shoulder Gridle when Viewed from the Front



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Her investigations revealed normal hemogram and biochemical parameters CPK was 345 units per litre, LDH-200 units per litre, SGOT and SGPT were normal. EMG was done which showed myopathic pattern (*Fig 3*).Muscle biopsy showed features of myopathy with segmental necrosis of muscle fibres with an occasional inflammatory infiltrate.

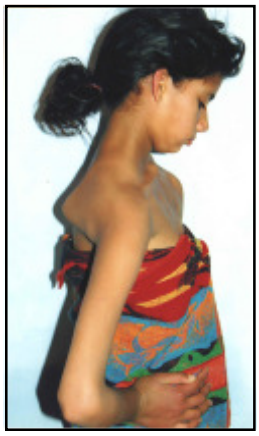

Fig. 2 Characteristic Appearance of Shoulder Gridle When Viewed from Lateral Side

Discussion

FSHD is a less common disorder with autosomal dominant inheritance (4). Our case did not have any family history and was sporadic in nature.The disease usually starts in adolescence but signs may be evident in children. Early in the disease, the muscle weakness may be asymmetrical and many of the patients with milder degrees of this form of dystrophy are unaware that they have the disease. This was true of nearly half of the very large series of patients described by Tyler and Stephens (5) in the utah Mormon population. At any point the disease may become virtually arrested. Nevertheless, 15-20% of patients eventually require a wheel chair (6) and very few may require ventilatory support as the disease progresses (7) . Other associated features have been noticed with the disease like mild sensorineural hearing loss and Vascular retinopathy (8). However, these findings are not consistent and it is not known how they relate the genetic abnormality. Cardiac arrythmias are rarely seen in this disease (FSHD).Over case of 14 years old female child with no family history and had weakness of face and arms and no associated abnormalities as they are rare as described in the literature (5).

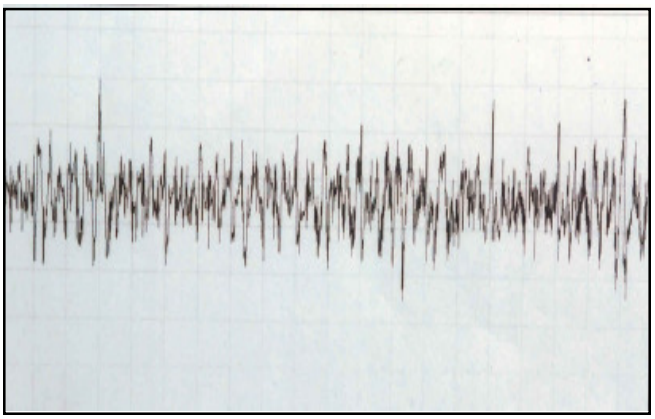

Fig. 3 Characteristic EMG Findings Showing Myopathic Pattern

Conclusion

FSHD is an uncommon disorder of muscles and should be suspected in children and not to be misdiagnosed as oculopharyngeal muscular dystrophy which has a late onset of 4th to 6th decade. There is no specific treatment available. Scapular stabilization procedures improve scapular winging. Need of the hour is proper counceling of the parents and patients regarding the disease.

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