Myopathy in the Fleet: An Atypical Case of Facioscapulohumeral Dystrophy Type 1

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INTRODUCTION

Facioscapulohumeral dystrophy (FSHD) is an autosomal dominant condition with variable penetrance predominantly affecting the facial and shoulder muscles. FSHD affects the face initially, with impaired smiling or whistling. Subsequently, patients develop scapular winging, humeral, truncal and lower extremity weakness.

CASE DESCRIPTION

A 26 year old male military servicemember presented with chronic right shoulder pain. At 19 years of age, he started weightlifting consistently but noticed his right pectoral muscle and trapezius muscles were getting thinner. He was seen by a shipboard physician who diagnosed him with “Poland syndrome”. He developed difficulty with pullups and pushups and right shoulder pain. MRI revealed labral tear and glenohumeral osteoarthritis. He had no family history of nerve or muscle disease. There was prominent right trapezius, rhomboid, and pectoralis atrophy without fasciculations or spasticity. He was unable to abduct his shoulder beyond 90 degrees. There was no facial weakness and the patient could readily whistle. EDX evaluation revealed right spinal accessory mononeuropathy and a right axonal CTS. An additional EDx study roughly 2 years later revealed myopathic processes in the right greater than left shoulder and thoracic paraspinals without myotonia. Genetic testing revealed a 4q35 deletion consistent with a diagnosis of FSHD1.

FIGURE 1: EMG

- **EMG 2.5 years prior to diagnosis:** Notable for a right spinal accessory mononeuropathy.
- **EMG 1.5 years prior to diagnosis:** Notable for a Myopathic process involving the right greater than left proximal shoulder and thorax muscles without evidence of myotonia.
- **EMG 1 year prior to diagnosis:** Notable for increased insertional activity and myopathic units in right pectoralis major, right trapezius. Increased insertional activity in the right biceps.
- **EMG Prior to Genetic Testing:** Notable for increased insertional activity observed in the right pectoralis major, right biceps brachii and right trapezius. Short, polyphasic units were seen in the right pectoralis major with a normal recruitment pattern. Intermixed pattern of large MUAPs and short polyphasic MUAPs is seen in the right trapezius muscle, with a normal recruitment pattern. Normal MUAPs and recruitment pattern is seen in right biceps brachii.
  - **Conclusion: Abnormal study due to:** Increased insertional activity in all muscles tested together with myopathic units seen on R pectoralis major and R trapezius muscle indicating ongoing irritable myopathy.

EMG Summary Table									
	Spontaneous					MUAP			Recruitment
Muscle	IA	Fib	PSW	Fasc	Other	Amp	Dur.	PPP	Pattern
R. Pectoralis major	Incr	1+	1+	None	CRDs	N	1-	1+	N
R. Biceps brachii	Incr	1+	1+	None	None	N	N	N	N
R. Trapezius	Incr	2+	2+	None	CRDs	N	N	1+	N

FIGURE 2: LABORATORY TESTING

	Site / Specimen		Units	Ref Range
Creatine Kinase	SERUM	639 (H)	U/L	(39-308)

FSHD-(FSHD1 and FSHD2) Detection of Abnormal Alleles with Interpretation (Final result) - collected:

4q35 deletion consistent with facioscapulohumeral dystrophy type 1 (FSHD1).

CONCLUSION

We report a patient with FSHD Type 1 who lacked the typical facial stigmata. His phenotype included asymmetric pectoral, trapezius and rhomboid atrophy without facial weakness. Roughly 90% of patients affected with FSHD have a 4q35 deletion. He had no family history of such disease, raising the question of a de novo germline mutation in this patient. This disease is important to recognize to ensure appropriate multidisciplinary evaluation of the extra-muscular manifestations it may manifest. This patient was referred for audiology, cardiology, ophthalmology and pulmonology multidisciplinary evaluation to rule out high frequency hearing loss, cardiac conduction abnormalities, retinal telangiectasias and restrictive lung diseases respectively.

In conclusion, consideration of neuromuscular and myopathic causes of scapular winging is necessary even in the absence of the stigmata of related diseases. Additionally, it is important to consider that shoulder pain may be the initial presentation of myopathic disease, even without a family history of myopathy.

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