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FASCIOSCAPULOHUMERAL MUSCULAR DYSTROPHY : A DIAGNOSTIC DILEMMA

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ABSTRACT

BACKGROUND - Facioscapulohumeral dystrophy (FSHD) is often cited as the third most common form of muscular dystrophy. However, supportive measures involving physical therapy and the use of orthotics may aid in improving function and mobility.

CASE SUMMARY – The patient is a 15-year-old right-handed male who presented with some right lower extremity weakness and atrophy. During the review of symptoms, the patient added difficulty lifting his arms overhead and a "crooked" smile to the list of complaints. The physical examination noted an asymmetric smile, both sided lateral scapular winging, and a steppage gait. Furthermore, atrophy of the right pectoralis major, trapezius, and gastrocnemius muscles were noted. In addition, atrophy of the bilateral tibialis anterior muscles was noted as well. Given that the results of the electrodiagnostic testing were consistent with a myopathic disorder, facioscapulohumeral dystrophy (FSHD) was suspected and genetic testing was ordered to confirm the diagnosis.

CONCLUSION - Individuals with FSHD should be seen at a frequency based on their disease severity, which for some will be frequent initially, and may include occupational and speech therapy in infantile onset forms of FSHD. Physical therapy and rehabilitation consultations can help establish appropriate exercise regimens and assistive devices that may enhance mobility and reduce the risk of falls in home environments.

KEYWORDS : facioscapulohumeral dystrophy(FSHD), Landouzy–Dejerine disease, muscular dystrophy, telomeric deletion disease, electrodiagnostic testing.

INTRODUCTION –

Facioscapulohumeral dystrophy (FSHD) is often cited as the third most common form of muscular dystrophy. So, it should be considered in patients with complaints of progressive weakness. We present the case of a male with facial, truncal, and leg weakness that initially sought medical attention for lower back pain. Electrodiagnostic testing revealed findings in the trapezius, Serratus anterior, biceps, triceps, pectoralis major, tibialis anterior, and gastrocnemius muscles consistent with a myopathic disorder. Subsequent genetic testing identified a FSHD allele size consistent with a FSHD deletion mutation. Therefore, confirming the diagnosis of FSHD. Unfortunately, no effective treatments currently exist for FSHD. However, supportive measures involving physical therapy and the use of orthotics may aid in improving function and mobility.

CASE REPORT-

The patient is a 15-year-old right-handed male who presented to his primary care provider with complaints of persistent lower back pain. After noticing little improvement of his symptoms with conservative management, including the use of nonsteroidal antiinflammatory drugs (NSAIDs), the patient was referred for further evaluation and electrodiagnostic testing. The majority of the primary care provider's work-up was unremarkable. An elevated creatine kinase (CK) level of 324 U/l was noted on routine serology. While describing the history of the present illness , the patient explained that he also noted some right lower extremity weakness and atrophy. Furthermore, the weakness made it difficult for him to walk properly at times. During the review of symptoms, the patient added difficulty lifting his arms overhead and a "crooked" smile to the list of complaints. The physical examination noted an asymmetric smile, both sided lateral scapular winging, and a steppage gait. Furthermore, atrophy of the right pectoralis major, trapezius, and gastrocnemius muscles were noted. In addition, atrophy of the bilateral tibialis anterior muscles were noted as well. (Figure 2,3,4). Manual muscle testing was 5/5 throughout the bilateral upper and lower extremities with the exception of the findings noted in Table 1. The remainder of the physical examination was unremarkable.

Electrodiagnostic testing of the upper and lower extremities resulted in a normal nerve conduction study (NCS). However, electromyography (EMG) did demonstrate findings consistent with a myopathic disorder. These results are summarized in Tables 2,3. The needle examination noted abnormal spontaneous activity in the form of fibrillation potentials and positive sharp waves (PSW) in various muscles including the right gastrocnemius and bilateral tibialis anterior muscles. Furthermore, short duration polyphasic potentials were noted in numerous muscles of the right periscapular region as well as of the bilateral lower extremities. Tracings obtained during the testing of the right tibialis anterior, such as that depicted in Fig. 1, are representative of these findings. Given that the results of the electrodiagnostic testing were consistent with a myopathic disorder, facioscapulohumeral dystrophy (FSHD) was suspected and genetic testing was ordered to confirm the diagnosis.

Genetic testing, performed at an outside facility, reported that the patient possesses a FSHD allele size measuring 35 kilobases (kb) that is consistent with a FSHD deletion mutation. However, the analysis also noted a possible benign translocation between the chromosome 4q35 and chromosome 10q26 loci that occurs in approximately 20% of the general population. Therefore, given that the genetic testing was only suggestive and not confirmatory for FSHD, the patient was referred to a neurologist specializing in the diagnosis and treatment of muscular dystrophy.

The neurologist requested that the patient don an ankle foot orthosis (AFO) in order to aid with ambulation given tibialis anterior weakness as well as ordering physical therapy in hopes of strengthening the affected musculature. In addition, it was recommended that the genetic testing be repeated at a second facility specializing in muscular dystrophy analysis. The results of the repeat genetic testing did confirm FSHD noting that the patient has a chromosome 4q35 deletion with restriction fragments smaller than 35 kb. Subsequently, the patient revisited with approximately 1 year after being diagnosed with FSHD. The patient noted improved ambulation with the use of the AFO. Furthermore, manual muscle testing noted significant improvement in the strength of the involved musculature. In addition, the CK level was 202 U/l at the time of revisit. Hence, 122 U/l less than the initial level measured by the primary care provider. Therefore, the patient was satisfied with his care and had taken the initiative to learn more about FSHD.

Table 1 Manual muscle testing

Muscle	Right	Left
Orbicularis oculi	4/5	4/5

Pectoralis major	4/5	5/5
Tibialis anterior	2/5	4/5
Peroneus longus	3/5	4/5
Extensor hallucis longus	3/5	4/5
Gastrocnemius	4/5	5/5
Hamstrings	4/5	4/5

Figure 1 Electromyographic tracing of(Rt) Tibialis Anterior

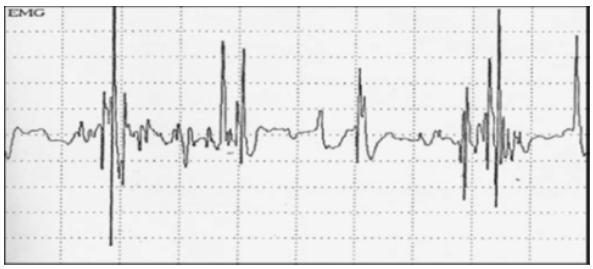

Table 2 Nerve Conduction Study

Nerve	Muscle	Lat 1	Lat 2	Lat2-Lat1	Amplitude	Temp (°c)
L Peroneal F	EDB	50.63		50.63		30.0
R Peroneal F	EDB	53.44		53.44		30.0
L Tibial F	AHB	49.53		51.09		30.7
R Tibial F	AHB	49.53		49.53		30.5

Table 3 Nerve Conduction Study: Sensory nerves

Nerve	Site	Onset latency (ms)	Peak latency (ms)	Amplitude	Duration (ms)	Seg name	Distance (cm)	Velocity (m/sec)
LS Peroneal	Ankle Lat leg	2.31	2.91	O-P (microV) 20.58	Neg 1.47	Lat leg-ankle	10.50	45.4
RS Peroneal	Ankle Lat leg	2.09	2.84	O-P (microV) 18.46	Neg -	Lat leg-ankle	11.00	52.5
L Sural	Lat malleolus Calf	2.09	2.88	O-P (microV) 20.28	Neg -	Calf - lat mal	10.50	50.1
R Sural	Lat malleolus Calf	1.88	2.64	O-P (microV) 20.64	Neg 1.38	Calf –lat mal	10.00	53.3

DISCUSSION-

French physicians, Louis Théophile Joseph Landouzy and Joseph Jules Dejerine, were the first to describe FSHD. This autosomal dominant disorder is clinically defined as progressive asymmetric muscular weakness typically of the face, scapular stabilizers, proximal arm, and leg. However, clinical presentations may vary. Uncini and his colleagues report of cases in which patients presented simply with asymmetrical atrophy of the calf muscles. Additionally, Van Der Kooi and his colleagues also present cases of leg weakness without any apparent involvement of facial muscles. Despite the various methods available to diagnose FSHD, no effective treatment is currently available for patients with the disease. Various studies examine the possible role of beta2-adrenergic agonists, such as albuterol, in treating FSHD. It is hypothesized that the use of such medications may exert anabolic effects on muscles and prevent atrophy after a variety of insults. Kissel and his colleagues explain that, although the use of albuterol does not improve overall strength or function in patients with FSHD, the medication may improve muscle mass. However, a Cochrane Database System Review of medical literature from 1966 to 2003 concludes that there is no evidence from randomized controlled trials to support any drug treatment for FSHD. Therefore, treatment often involves supportive measures to aid in improving the patient's function. Orthotics, such as an AFO, may be used to aid in improving ambulation. In addition, exercise and physical therapy may result in less fatigue and increased mobility. Olsen and his colleagues note that a 12-week low-impact aerobic program improves maximal oxygen intake and workload without further damaging muscles. Future treatments, however, may involve the use of stem cells in order to possibly treat FSHD at the cellular level. Morosetti and his colleagues discuss the possible use of autologous mesoangioblasts from the patient's own healthy muscle to limit damage within the diseased musculature. Vilquin and his colleagues offer a similar suggestion as they discuss the possible use of autologous myoblasts in order to treat FSHD. Finally, experts highly recommend genetic counseling for individuals with FSHD who are planning to have children. \

CONCLUSION - In the pre-molecular era, the diagnosis and counselling of FSHD families was entirely based on clinical evidence(1). Over the years, DNA testing of the D4Z4 locus and flanking polymorphisms has been considered highly sensitive and specific and extensively used to diagnose FSHD(1). Standards of care and management of facioscapulohumeral muscular dystrophy were agreed upon at the 171st ENMC International Workshop(2).The Physical therapy and rehabilitation consisted of Consultation with a physical therapist, Establishment of follow-up frequency is important at the time of diagnosis. Individuals with FSHD should be seen at a frequency based on their disease severity, which for some will be frequent initially, and may include occupational and speech therapy in infantile onset forms of FSHD. For others with mild involvement, annual visits would be appropriate. Physical therapy and rehabilitation consultations can help establish appropriate exercise regimens and assistive devices that may enhance mobility and reduce the risk of falls in home environments. The exercise programme consisted of Exercise with moderate weights, and is not detrimental to individuals with FSHD. Aerobic training (when possible) has been beneficial to affected individuals . Any type of exercise regimen should be instituted under the guidance of a physical therapist and personalized according to the individual's disease symptoms, age, and cardiovascular status.(2) Training sessions consists of a 30-minute aerobic exercise period with a warming-up and cooling-down period of 5 and 3 minutes, respectively.(3)

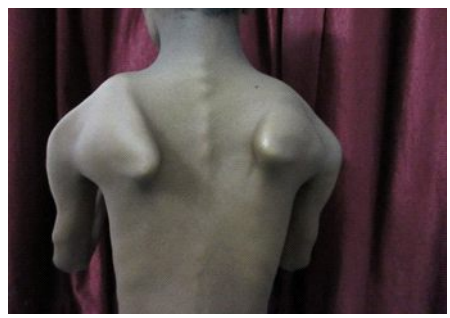

FIGURE 2: SHOWING BOTH SIDED LATERAL SCAPULAR WINGING

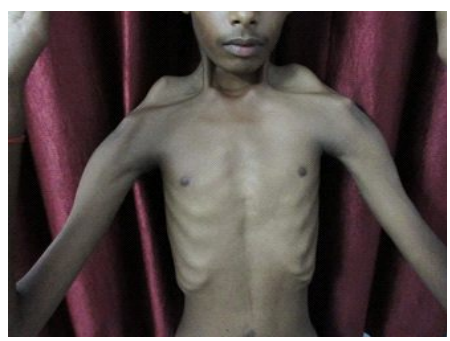

FIGURE 3: SHOWING ATROPHY OF THE RIGHT PECTORALIS MAJOR, TRAPEZIUS MUSCLES WITH INABILITY TO PERFORM OVERHEAD ABDUCTION

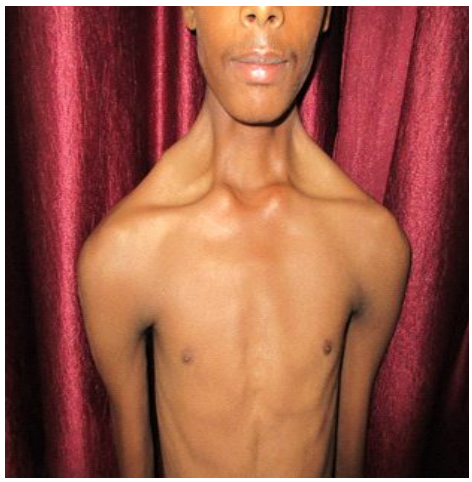

FIGURE 4: SHOWING "CROOKED SMILE" AND WASTING OF ORBICULARIS ORIS

Conflict of interest - The authors declare no conflict of interest whatsoever arising out of the publication of this manuscript.

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