A case of fascioscapulohumeral muscular dystrophy misdiagnosed as Becker’s muscular dystrophy for 20 years

VESPER FE MARIE LLANEZA RAMOS, PARIWAT THAISETTHAWATKUL

Neurological Sciences, University of Nebraska Medical Center, 982045 Nebraska Medical Center, Omaha, NE 68198-2045, USA

Address correspondence to: V. F. M. L. Ramos. Tel: (+1) 402-889-2284; Fax: (+1) 402-559-3341. Email: vesper.ramos@yahoo.com; ramosv@unmc.edu

Abstract

A 60-year-old man diagnosed clinically with Becker’s muscular dystrophy 20 years ago by another physician presented with gradually progressive proximal muscle weakness since teenage years. Family history revealed a strong paternal familial inheritance pattern of similar distribution of weakness—face, forearm flexion, knee extension and foot dorsiflexion. Work-ups revealed B12 deficiency and allele 1 deletion in fascioscapulohumeral muscular dystrophy (FSHD) DNA testing. FSHD is the third most common muscular dystrophy. Clinical diagnosis is made from the distinctive pattern of weakness, autosomal-dominant inheritance, and confirmed by genetic testing. This case strongly demonstrates the importance of a thorough and careful clinical evaluation even in a case with a long-standing diagnosis.

Keywords: Fascioscapulohumeral muscular dystrophy, progressive weakness, familial weakness, elderly

Case report

A 60-year-old man with type 2 diabetes mellitus (DM) presented with gradually progressive muscle weakness, starting proximally, since teenage years. He was diagnosed 20 years ago by another physician with Becker’s muscular dystrophy. At the time of his diagnosis, creatine phosphokinase (CPK) levels were normal on two different occasions. Genetic testing was never performed. He started using a leg brace 4 years before. He was wheelchair-bound for the past year.

He reported similar weakness in his father, now 86 years of age, also wheelchair-bound. Weakness of similar distribution was found in four paternal uncles and grandmother. He also had a 56-year-old brother, who used a cane, but attributed weakness to a motor vehicle accident.

Physical exam revealed bilateral facial weakness, most prominent on eye closure and smiling, atrophy of the trapezius and pectoralis major muscles and an elevated scapula with internal rotation of the right arm. Motor weakness was seen in forearm flexion, knee extension and foot dorsiflexion. Ankle reflexes were absent.

Complete blood count and metabolic profile were normal. CPK was 35 units per litre. Vitamin B12 was decreased at 150 pg/ml (111 pmol/l). Antinuclear antibody panel, rheumatoid factor, HIV, Lyme antibody and serum angiotensin-converting enzyme levels were non-reactive. Electrophysiological studies revealed chronic active myopathy and chronic axonal sensorimotor polyneuropathy. Fascioscapulohumeral muscle dystrophy (FSHD) DNA testing showed allele 1 deletion.

Discussion

In this case, the diagnosis of Becker’s muscular dystrophy, an X-linked disorder, raises doubt because of evident
father-to-son transmission, which is characteristic of an autosomal dominant disorder. Normal CPK levels when the patient was already symptomatic but remaining ambulatory also argues against Becker’s.

FSHD is the third most common muscular dystrophy, with a prevalence of 1/20,000 [1]. The name was coined by George Padberg in 1980 in a retrospective review of 107 cases. FSHD is also known as Landouzy-Dejerine muscular dystrophy, as first described by Louis Theophile Joseph Landouzy and Joseph Jules Dejerine in 1885 [2–4].

Clinical features include progression of muscle weakness, first in the face, then scapular stabilisers and then proximal arm, which provides the rationale for its name. Orbicularis oculi and orbicularis oris are commonly affected. Oculomotor, laryngeal and pharyngeal muscles are typically spared. Scapular elevation with winging during shoulder flexion or abduction is common, as is internal rotation of the humerus. Atrophy of the pectoralis major muscle is an early finding [5]. Hamstrings, gastrocnemius and tibialis anterior are the most involved in the lower extremities.

Diagnosis can be made with relative certainty from the distinctive pattern of weakness plus autosomal-dominant family history. It is confirmed with molecular diagnosis (sensitivity and specificity of 95%) with deletion of D4Z4 repeats in one copy of chromosome 4q35 [5]. Residual repeat number is inversely related to the age of onset and severity of the disease. Mouse transfection studies recently demonstrated a genetic model for FSHD, with specific single nucleotide polymorphisms in the region distal to the last D4Z4 repeats [6].

There is no known cure. Corticosteroids, albuterol, creatine monohydrate and myostatin have not been beneficial. Aerobic exercise improve strength and endurance but not fatigue symptoms [7].

Even in the light of obvious diagnoses, a thorough work-up is necessary so that confounding correctable factors are not missed. In this case, B12 deficiency, and not FSHD, could explain the findings of polyneuropathy on the electrophysiological study.

A careful clinical evaluation with thorough and thoughtful diagnostic work-ups is still necessary in evaluating patients with long-standing diagnoses.

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**Key points**

- Careful clinical evaluation, including family history, is warranted even in the evaluation of the elderly patient with a long-standing diagnosis. Genetic diseases can be seen in the elderly.
- A thorough and thoughtful diagnostic work-up, guided by clinical acumen is necessary in the work-up of the elderly patient with a suspected genetic disease. Confounding and/or correctable factors must be considered.
- Genetic testing offers confirmatory diagnosis, with high sensitivity and specificity, for many muscular dystrophies and other genetic illnesses. Requesting these tests must be guided by the clinical evaluation.

**Conflicts of interest**

None declared.

**References**


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