Facial paresis as the first sign in atypical facioscapulohumeral muscular dystrophy

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ABSTRACT

Background: Facioscapulohumeral muscular dystrophy (FSHD) is the one of the most common types of muscular dystrophy. We present a retrospective case description of a patient with late-onset, atypical FSHD and provide an overview of the clinical history, physical exam findings, diagnosis and treatment of FSHD.  
Main findings: A 71-year old male with subjective facial weakness and dysarthria presented initially without physical exam findings of paresis and normal diagnostic lab work. Over time, unilateral incomplete facial paresis appeared on physical exam, as well as mild scapular winging.  
Conclusion: FSHD classically presents with weakness in muscles of the face, shoulder/upper arms, and proximal lower extremities. Diagnosis is challenging and requires a multidisciplinary approach, due to high variability in clinical presentation and timing of symptoms. A supplementary video is provided, demonstrating unilateral midfacial and lip paresis in a 71-year-old male.

1. Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is the one of the most common types of muscular dystrophy, affecting 1 out of 15–20,000 people [1,2]. The typical presentation is characterized by its name: weakness of the facial, shoulder, and upper arm muscles. However, due to variable timing and severity of myopathy, the clinical picture makes diagnosis of FSHD is challenging. Diagnosis and management of FSHD require a multidisciplinary approach. The Washington University School of Medicine Institutional Review Board exempted this research. Patient consent for publication was obtained.

2. Case report

A 71-year-old male initially presented to a neurologist with dysarthria. Extensive workup (routine labs, vitamin B12, erythrocyte sedimentation rate, and creatine kinase) were within normal limits. Antinuclear antibody and acetylcholine receptor-binding antibody (to rule out myasthenia gravis) were negative. MRI/MRA Brain and MRI Head and Neck were negative for intracranial abnormality, or neck masses and facial nerve abnormalities, respectively. Otologic exam and audiometry were normal and he did not complain of hearing loss. He presented to the Facial Plastic and Reconstructive Surgery clinic 17 months later, with new midfacial weakness, oral dysphagia, and worsening dysarthria. Physical exam at this time showed right midfacial and lip paresis (Fig. 1, see also Supplemental Video 1, demonstrating right midfacial and lip paresis). The patient then followed up with a different neurologist, who noted that he had mild winging of his left scapula. Electromyography (EMG) demonstrated slowed conduction velocities and small compound muscle action potentials. Given this patient’s late-onset, asynchronous, and asymmetric facial weakness, left scapular winging, and non-specific EMG results, he was diagnosed with atypical FSHD.

3. Discussion

FSHD is the third-most common muscular dystrophy, after Duchenne muscular dystrophy and myotonic dystrophy [1]. Diagnosis is challenging and requires a multidisciplinary approach. The presented case, with symptoms of late-onset progressive facial weakness and scapular winging, underwent extensive workup, including management by Neurology, Neurotology and Facial Plastic Surgery specialists. FSHD classically presents with asymmetric and progressive muscle...
weakness, initially of the face (periocular or perioral weakness), shoulder/upper arms (scapular winging and upper arm weakness with forearm sparing), and proximal lower extremities [3]. Symptoms usually start in the first or second decade of life. Myopathy occurs proximally to distally, and approximately 20% lose the ability to ambulate, becoming wheelchair-bound [3]. However, FSHD can occur at any time in life; it is often asymmetric (affecting different sides of the body) and asynchronous in timing. Extra-muscular manifestations are rare: retinal vascular changes, pulmonary complications from respiratory muscle weakness, and hearing loss [2,3]. People with FSHD have a low quality of life, similar to other types of muscular dystrophy.

Apart from clinical and family history (30% have no family history), genetic testing is sensitive and specific. Ninety-five percent of cases have FSHD1: autosomal dominant deletion in repeats in chromosome 4q (D4Z4). FSHD2, due to reduced methylation of this same gene region, occurs in five percent [2]. MRI, muscle biopsy, and EMG are all relatively non-specific [3–5]. Clinicians should have a low threshold to screen for extra-muscular manifestations (audiometry, pulmonary function tests, ophthalmology exam). Treatment involves rehabilitation and pain control. Currently, no FDA-approved treatments exist; there are few clinical trials of gene therapies [3,4].

FSHD is a rare, systemic disease that can manifest with facial paresis. Atypical presentations make clinical diagnosis challenging. The American Academy of Neurology (AAN) and the American Association of Neuromuscular and Electrodiagnostic Medicine (AANEM) have provided guidelines for diagnosis (Fig. 2) and treatment of FSHD, which may be found online at https://www.aan.com/Guidelines. Although otolaryngologists are not typically making the diagnosis of FSHD, they are often involved in care of patients with cranial neuropathies. This case underscores the importance of early recognition and prompt involvement of neurology in a multidisciplinary approach.

Fig. 1. Right facial paresis in a patient with atypical FSHD. The right nasolabial fold is slightly effaced. There is limited protrusion of the right lower lip.

### Author contributions

Nneoma S. Wamkpah, conceptualization, methodology, writing – original draft; John J. Chi, conceptualization, methodology, writing – original draft.

### Conflicts of interest

None.

### Sponsorships

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### Ethical statement

The Washington University School of Medicine Institutional Review Board exempted this research. Patient consent for publication was obtained.
Declaration of competing interest

The authors (Nneoma S. Wamkpah, MD; John J. Chi, MD MPHS) have no conflicts of interest.

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