A Great Athlete with Muscular Weakness

Rim Bourguiba, Marion Masingue, Pierre-Jean Marc, Thimothée Lenglet, Claude Bachmeyer

A great athlete with muscular weakness

Rim Bourguiba MD , Marion Masingue MD , Pierre-Jean Marc MD ,
Thimothée Langlet MD , Claude Bachmeyer MD

Please cite this article as: Rim Bourguiba MD , Marion Masingue MD , Pierre-Jean Marc MD , Thimothée Langlet MD , Claude Bachmeyer MD , A great athlete with muscular weakness, *The American Journal of Medicine* (2020), doi: https://doi.org/10.1016/j.amjmed.2020.07.030

Title: A great athlete with muscular weakness

Authors:

Rim Bourguiba, MD[a]

Marion Masingue, MD[b]

Pierre-Jean Marc, MD[c]

Thimothée Langlet, MD[d]

Claude Bachmeyer, MD[a]

Author's affiliation

[a]Department of Internal medicine, Assistance publique-Hôpitaux de Paris, Hopital Tenon, 4 Rue de la Chine 75020

[b]Reference Center for Neuromuscular Diseases Nord-Est-Ile de France, Institute of Myology, CHU Pitié Salpetrière, Assistance Publique-Hôpitaux de Paris, Paris, France.

[c]Department of Genetics, Hôpital Cochin, Assistance publique-Hôpitaux de Paris, Paris,France

[d]Department of Clinical Neurophysiology, Hôpitaux universitaires Pitié-Salpêtrière Charles Foix, Assistance Publique-Hôpitaux de Paris (APHP), Paris, France

Funding: None

Conflict of interest: None

Authorship : All authors had access to the data and a role in writing this manuscript.

Corresponding

Rim Bourguiba

Department of internal medicine

Hopital Tenon, 4 Rue de la Chine

75020

Tel +33156016077

Mail rim.bourguiba@aphp.fr

*To the Editor***:**

A 21-year-old Romanian man presented to our clinic for leanness assessment evolving for several years. He had been adopted at the age of 3 years old hence, no familial history was available. His medical history was notable for dyspraxia, dyslexia, bilateral cryptorchidism and scoliosis in infancy. He practiced water polo, swimming and martial arts, but in recent years, amyotrophy and weakness of both arms had developed (Figure, a) without dyspnea or swallowing troubles. Search for an underlying disease including malabsorption, endocrinopathy, infections, cancer, and anorexia nervosa had been ruled out previously by many specialists. On physical examination the patient was healthy, with a body mass index of 17.5. Physical examination showed distal wasting of the deltoid muscle with a normal proximal part, wasting of the humeral muscle, and bilateral scapular winging (Figure, a,b) highly suggestive of facioscapulohumeral muscular dystrophy (FSHD). Muscle testing revealed bilateral weakness of deltoid and biceps muscles (4/5 MRC scale) as well as abdominal muscles and hip abductors on both sides. Orbicular muscles (eyes and lips) were also weak. No gait disturbance was noted. Routine blood examination was unremarkable but

serum creatine phosphokinase level of 559 UI (<195UI), with negative dot myositis. Electroneuromyography (EMG) showed normal nerve conduction study and asymmetric myopathic features predominately in the facial and scapulohumeral muscles. Mild diaphragmatic insufficiency was diagnosed (vital capacity 77%). Genetic testing confirmed FSHD type 1. D4Z4 contraction using southern blot showed loss of microsatellite repeated units in the subtelomeric region of chromosome 4.

Discussion:

We report here a patient with typical clinical features of FSHD in whom the diagnosis was established late. Indeed, this disease is one of the commonest muscular dystrophy in adults with autosomal dominant inheritance. Clinical features include weakness of facial muscles and shoulder girdle musculature, resulting in scapular winging with an asymmetric muscle involvement, but also trunk and legs, and a positive family history.[1-3] There is no cardiac involvement, but respiratory muscles may be affected. EMG features and biopsy are often nonspecific.[2] Two genetically forms of FSHD are described: around 90% to 95% of patients have FSHD type 1, other patients have FSHD type 2. Patients with FSHD1 have a loss of D4Z4 macrosatellite repeated unit in the subtelomeric region of the long arm of chromosome 4q 2. Patients with FSHD2 have a mutation in the SMCHD1 gene (chromosome 18). Consensus criteria suggest screening for D4Z4 contraction using southern blot in patient with characteristic clinical presentation.[3]

To date, there is no specific treatment for FSHD and only supportive care is proposed with muscle stretching and exercise, assessing pulmonary and cardiac function with a baseline eye examination.[3] Some authors suggest scapular fixation can improve shoulder function.[4] As FSHD1 is a genetic disorder, use of antisense oligonucleotide therapy to target

DUX4 was proposed for FSHD but not actually tested on human.[5]

Physicians should be aware of subtle signs and symptoms of FSDH in order to allow an early diagnosis (confirmed on genetic testing) and appropriate management without needing multiple diagnostic procedures.

References

1. Statland J, Tawil R. Facioscapulohumeral Muscular Dystrophy. Neurologic Clinics. 2014;32(3):721-728.

2. van der Maarel SM, Tawil R, Tapscott SJ. Facioscapulohumeral muscular dystrophy and DUX4: breaking the silence. Trends in Molecular Medicine. 2011;17(5):252-258.

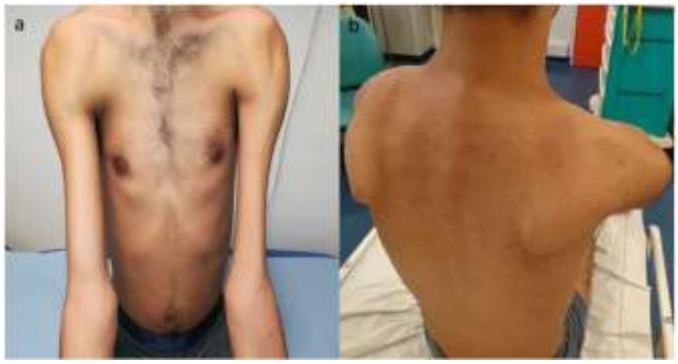
3. Tawil R, Kissel JT, Heatwole C, Pandya S, Gronseth G, Benatar M. Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: Report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology. 2015;85(4):357-364

4. Orrell RW, Copeland S, Rose MR. Scapular fixation in muscular dystrophy. Cochrane Neuromuscular Group, ed. Cochrane Database of Systematic Reviews. Published online January 20, 2010.

5. Statland JM, Tawil R. Facioscapulohumeral Muscular Dystrophy: CONTINUUM:

Lifelong Learning in Neurology. 2016;22(6):1916-1931.

Legend of figures



Figure

A: Distal wasting of the deltoid muscle with normal proximal part and wasting of the humeral muscle

B: Scapular winging and amyotrophy of upper limb