

# A rare case of facioscapulohumeral muscular dystrophy and myasthenia gravis

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## Keywords

Facioscapulohumeral muscular Dystrophy, Myasthenia Gravis, Myopathy, Iran

## Abstract

Facioscapulohumeral muscular dystrophy (FSHD) is a common inherited muscular dystrophy presented clinically with slowly progressive weakness and wasting of facial and limb muscles and rare bulbar muscle involvement. We present herein a 70-year-old man who was a known case of FSHD with complaint of 15-day history of progressive difficulty in chewing and dysarthria and was found to have myasthenia gravis. Related literatures have been also reviewed.

## Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is a common inherited muscular dystrophy presented clinically with slowly progressive weakness and wasting of facial and limb muscles and rare bulbar muscle involvement.<sup>1</sup> We present herein a known case of FSHD presented with recent onset of severe bulbar symptoms and was found to have myasthenia gravis (MG) based on electrodiagnostic study, elevated level of acetylcholine receptor antibody and dramatic improvement with choline esterase inhibitor agents.

Concomitant occurrence of FSHD and MG is rare according to the medical literature. Clinical presentation, electrodiagnostic and pathologic findings of this patient are described.

## Patient

A 70-year-old man presented to our department with complaint of 15-day history of progressive difficulty in chewing and dysarthria. He had a 50-year history of slowly progressive asymmetrical weakness of proximal upper limb muscles.

Examinations revealed reduction in forces of bilateral orbicularis oculi muscles, weakness and wasting of bilateral triceps muscles especially on right side and bilateral winging of scapula more prominent on right side. The legs and pelvic girdle muscles had normal forces. The reflexes were somewhat depressed throughout. Sensation and coordination were normal. The patient's gait was normal.

Serum creatine kinase values and other laboratory data were normal. Computerized tomography of the thorax was normal. Nerve conduction studies showed decrement response on repetitive nerve stimulation. Concentric needle electromyography showed myopathic changes especially in proximal limb muscles. Edrophonium test was performed and dysarthria and chewing difficulty showed dramatic improvement but had no effect on limb weakness.

Serum anti-acetylcholine receptor (AChR) antibody level was markedly elevated. Muscle biopsy showed myopathic changes with invariability in muscle fiber size, intramuscular infiltration of chronic inflammatory cells, mostly lymphocytes, few hyaline fibers and prominent fat infiltration. Subsequent genetic analysis confirmed the diagnosis of FSHD. Our patient's bulbar symptoms showed dramatic improvement following administration of choline-esterase inhibitor agents.

## Discussion

FSHD is the third most common inherited muscular dystrophy. It presents clinically with slowly progressive weakness and wasting of facial and shoulder girdle muscles and sometimes involvement of lower extremities.<sup>1</sup> Bulbar muscle is typically spared in FSHD. The clinical severity is wide ranging from asymptomatic individuals to wheel-chair dependent patients.<sup>2</sup> Any unusual changes in course of disease or development of unusual symptoms should raise the possibility of concomitant disease.

The coexistence of FSHD and MG has also been described previously.<sup>3-5</sup> Sansone et al.<sup>3</sup> reported a 69-year-old known case of FSHD who presented with sudden deterioration of limb weakness and development of bulbar symptoms and was found to have MG based on repetitive nerve stimulation, elevated level of acetylcholine receptor-binding antibody and dramatic improvement following immunomodulator administration. Sakuma et al.<sup>4</sup> reported a 50-year-old man with a 35 year history of FSHD who presented with acute progressive weakness of lower extremities three weeks prior to admission. The patient was found to have MG based on decrement response on repetitive nerve stimulation, elevated level of acetylcholine receptor-binding antibody and improvement after thymectomy and administration of corticosteroid. In another report,

McGonigal et al.<sup>5</sup> presented a 56-year old newly diagnosed myasthenic patient who found to have 40-year history of progressive foot drop and was found to have FSHD based on muscle biopsy and genetic analysis.

Although our case is a rare coexistence of FSHD and MG, low prevalence of both diseases, may raise the possibility of the presence of other etiologies. Theoretically, it is related to breaking of immune tolerance to AChRs as a result of muscle fiber degeneration. Patients with genetic myopathies may occasionally develop antibodies to AChR.<sup>6</sup> While these antibodies may not have pathogenic effects, their production is likely to be a consequence of sensitization to AChR secondary to muscle fiber damage, rather than through an immune process in thymus. Another theory could be a role of immune mechanism in the pathogenesis of FSHD. Muscle histopathological examination in some cases of FSHD shows inflammatory changes, disproportionate to muscle fiber necrosis but the presence of mononuclear infiltration does not affect disease progression and the patients do not benefit from prednisone treatment.<sup>7</sup>

## Conclusion

To the best of our knowledge, there are limited reports of concomitant occurrence of MG and FSHD in literatures. Although the association of MG with FSHD in our patient could be an incidental finding, it may raise the possibility of innate immune response, i.e. autoinflammation in development of AChR antibodies in genetic myopathies or it may suggest immune mechanisms in pathogenesis of FSHD. Moreover, our experience may warrant concomitant neuromuscular disorder in patients with unusual symptoms of FSHD.

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