Case Report

Myasthenia Gravis Coexisting with Myotonic Dystrophy

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Abstract

Myasthenia gravis and myotonic dystrophy do not usually coexist; however, we present a rare case where both conditions coexisted. Herein, we describe a 34-year-old woman who presented with symptoms of myasthenia gravis with coexisting myotonic dystrophy. She complained of limb weakness, difficulty in chewing and swallowing, and ptosis. She also had myotonia. The patient’s brother also had similar symptoms. Myasthenia gravis was confirmed by elevated acetylcholine receptor antibodies and neostigmine test, and myotonic dystrophy was confirmed by genetic studies. She was started on pyridostigmine and steroids after which the symptoms improved.

Myotonic dystrophy is very rarely associated with myasthenia gravis; thus, it is important to be attentive for features of myasthenia gravis in patients with myotonic dystrophy.

INTRODUCTION

Myasthenia gravis is an autoimmune disease characterized by weakness in certain muscle groups. It may begin at any age from infancy to very old age. Patients seek medical attention for specific muscle weakness that typically worsens with activity. There is fluctuation of weakness and symptoms become worse as the day progresses.

Myotonic dystrophy is a muscle disorder characterized by weakness, muscle wasting, and myotonia. It is autosomal dominant in inheritance and has an incidence of 1 per 8000 live births. Myotonic dystrophy type 1 is caused by mutations in the DMPK gene located on chromosome 19q13.3, and has a predilection for the distal muscles, whereas, myotonic dystrophy type 2 has a predilection for the proximal muscles.

CASE PRESENTATION

A 40-year-old woman presented at our hospital with difficulty in getting up from the squatting position and difficulty in lifting objects overhead. She also had difficulty chewing and swallowing. There was history of ptosis but there was no history of diplopia. Her symptoms started 3 years ago and were progressive. There was a diurnal variation of limb weakness and ptosis. There was a family history of symptoms with her younger brother also having ptosis and generalized weakness. He also reported a diurnal variation of symptoms. They were the only people affected in the family (Figure 1).

Neurological examination demonstrated bilateral ptosis and bilateral facial weakness. Eye movements were normal. There was generalized weakness with involvement of the upper and lower-limb proximal and distal muscles. Her reflexes were sluggish and the Babinski sign was negative. Sensory examination was normal. We also observed percussion as well as grip myotonia of the hand muscles.

Her complete blood count, erythrocyte sedimentation rate, and serum chemistry were normal. Her acetylcholine-receptor antibody titer was elevated (1.95 nmol/L). Needle electromyography (EMG) showed myotonic discharges, short duration, and small amplitude motor-unit action potential (MUAP) with early recruitment (Figure 2). We also did Neostigmine test on the patient. We observed that there was improvement in ptosis after 1.5 mg of neostigmine (Figure 3A). Computed tomography of the thorax ruled out thymoma. Echocardiography was normal.

Ophthalmological evaluation was also normal and there was no cataract.

Her brother also underwent thorough neurological examination. He had frontal baldness, ptosis, generalized weakness, and grip myotonia (Figure 3 B). However, there was no diurnal variation of symptoms. Repeated nerve stimulation did not show any decremental response. EMG showed myotonic discharges and myopathic pattern of MUAP and recruitment.

We confirmed myotonic dystrophy via genetic studies. CTG trinucleotide repeat expansion in the 3’ untranslated region of DMPK (which maps to 19q13.3) showed only one allele in the normal range. Triple-repeat primed polymerase chain reaction showed the presence of an expanded allele.

The patient was started on pyridostigmine 60 mg three times daily. **Figure 2** EMG of patient. **Figure 3** A. Before (left) and after (right) neostigmine test, B. Patient’s brother.
times daily, and steroids. There was significant improvement in the ptosis and limb weakness. Repeat Acetyl choline receptor antibody done after 1 year was undetectable.

DISCUSSION

Myotonic dystrophy is very rarely associated with myasthenia gravis. To date, only 9 cases have been reported with coexisting myotonic dystrophy and thymoma [1-10]; 2 cases were of both thymoma and myasthenia gravis, and 3 cases were of myotonic dystrophy and myasthenia gravis [11-13]. A case reported a 34 year old patient with myasthenia and myotonic dystrophy type 1 who also had thymoma. His myasthenic symptoms improved after thymectomy, steroids and pyridostigmine [10].

In the patient reported, evidence of myasthenia consisted of diurnal variation of weakness, abnormal decremental response on repeated nerve stimulation, and improvement with parenteral neostigmine. Evidence of myotonic dystrophy included voluntary and percussion myotonia, needle EMG showing myotonia and myopathic changes, and genetic studies confirming myotonic dystrophy type 1.

Myasthenia gravis and myotonia were once considered pharmacologic opposites. With Walker’s discovery that pyridostigmine alleviates the muscular weakness of myasthenia gravis and Wolf’s finding that quinine helps treat myotonic dystrophy [14] pathophysiology of these disorders. They found that neostigmine, which improved myasthenia, worsened myotonia in both myotonic dystrophy and myotonia congenita. Conversely, quinine was effective in diminishing myotonia but aggravated myasthenia. They concluded that myotonia is due to the accumulation of acetylcholine or an insufficient concentration of cholinesterase at the motor endplate.

Our diagnosis of myasthenia gravis was based on elevated acetylcholine receptor antibodies which is specific for myasthenia gravis. There is a possibility of myasthenia gravis and myotonic dystrophy coexisting. Such patients need to be identified to give adequate treatment.

REFERENCES