CASE REPORT
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A Patient with Genetically Confirmed Myoclonus-Dystonia Responded to Anticholinergic Treatment and Improved Spontaneously

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Background The various medical treatments applied to myoclonus-dystonia patients with a mutation of the ε-sarcoglycan gene (*SGCE*) have not been beneficial in most cases. Most patients experience progressive deterioration or static clinical courses, with only rare cases of spontaneous remission.

Case Report A 19-year-old girl presented with a 14-year history of myoclonus and dystonia that severely affected her left arm, neck, and trunk. Genetic studies showed a mutation in *SGCE* [deletion in exon 6 (c.771_772delAT, Cys258X)]. Both myoclonus and dystonia responded to anticholinergic treatment for 7 years and improved spontaneously.

Conclusions The possibility of spontaneous improvement should be kept in mind when considering the therapeutic strategy in myoclonus-dystonia patients, especially when contemplating deep-brain stimulation.
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Key Words myoclonus, dystonia, anticholinergics, spontaneous remission.

Introduction

Myoclonus-dystonia (MD) is a familial disorder with autosomal dominant inheritance. It is associated with a mutation of the ε-sarcoglycan gene (*SGCE*) and usually occurs during childhood. MD is a mixture of myoclonus and dystonia that mainly affects the proximal parts of the limbs and the trunk.[1,2] About 80% of patients with genetically confirmed MD respond dramatically to alcohol ingestion, but most respond poorly to medical treatment.[1,2]

We describe a female patient who developed MD in association with an *SGCE* mutation. Both myoclonus and dystonia responded to anticholinergic treatment for 7 years and thereafter improved spontaneously.

Case Report

A 19-year-old girl visited our clinic due to generalized jerks and dystonia. Her birth, perinatal, growth, and development histories were unremarkable. She was a single child of nonconsanguineous parents. Her 51-year-old father had experienced fine, jerky, postural tremor of both hands for 40 years, but the tremor did not disturb his activities of daily living. At the age of 5 years, the patient's parents noticed jerks of their daughter's left foot. When she started to walk and run, she frequently fell due to twisting of the left foot. The jerks and dystonia progressively worsened up to the age of 13 years, spreading to involve the neck, trunk, and all four limbs. Thereafter, her symptoms stabilized for the following 6 years. The jerks and dystonia were aggravated by emotional stress and disappeared while she was sleeping. Her eye movements were normal. Detailed cognitive function tests revealed no abnormalities. She exhibited retrocollis and backward and right-side tilting of the trunk as well as generalized jerks and dystonia that severely affected the left arm (Video Segment 1). Her left arm took a dystonic posture

with flexion at the elbow, wrist, and metacarpophalangeal joints. She had no dysarthria or dysphagia. The dyskinesia improved markedly with the ingestion of small amounts of alcohol. Her motor and sensory function tests were normal. Deep-tendon reflexes were normoactive bilaterally, and plantar reflexes were flexor bilaterally. Cerebellar function tests were normal. Routine laboratory tests, measurements of serum ceruloplasmin, serum creatine kinase, and serum lactate levels were all normal. Her EEG, median somatosensory evoked potential studies, and brain MRI were normal. DNA analyses of the patient and her father showed the same mutation in the SGCE gene [deletion in exon 6 (c.771_772delAT, Cys258X)]. Genetic studies of the mutation of *DTY1* and *GCH-1* were negative. She did not respond to levodopa/carbidopa (100/25 mg t.i.d.) or clonazepam (0.5 mg t.i.d.). Her dystonia and myoclonus responded markedly to trihexiphenidyl (2 mg t.i.d.). She stopped taking the medication many times, which resulted in recurrence of the dystonia and myoclonus. When trihexiphenidyl was discontinued at 7 years after the initiation of treatment (Video Segment 2), neither the myoclonus nor dystonia recurred, and the spontaneous remission persisted for more than 1year.

Discussion

The patient had some clinical features that are atypical for MD, such as dystonia starting from the distal part of the leg, markedly asymmetric myoclonus and dystonia, good response to low-dose anticholinergic treatment, and spontaneous remission. However, her other clinical features (onset during childhood, dyskinesia consisting of dystonia and myoclonus, predominant involvement of the neck and trunk, and dramatic response to alcohol ingestion) were compatible with MD.[1,2] The diagnosis was confirmed by a genetic study showing an *SGCE* mutation [deletion in exon 6 (c.771_772delAT, Cys258X)].[2,3]

The various medical treatments applied to MD patients have not been beneficial in most cases.[1,2] Responsiveness to levodopa has been described among reports of patients with genetically confirmed MD.[2,4] In addition, some patients also benefit from anticholinergic drugs;[1,2,4] however, they show a modest improvement that is frequently undermined by intolerable side effects. More favorable responses to anticholinergic treatment have been reported in several patients diagnosed with essential myoclonus or myoclonic dystonia, but their clinical diagnoses were not confirmed by genetic studies.[5-7]

Most patients with *SGCE* mutations exhibit progressive deterioration or static clinical courses. However, although spontaneous remission of dystonia may occur in about 20% of patients with MD during childhood or adolescence, the rate of spontaneous remission of myoclonus is only about 5%.[2,3] Interestingly, our patient showed spontaneous remission of jerks and dystonia in her 20s. Therefore, although most patients with MD do not respond well to medical treatment, the present case suggests that deep-brain stimulation should be delayed until patients are in their mid-20s.

Video Legends

Segment 1. A 19-year-old girl shows generalized jerks and dystonia severely affecting the left arm. Her left arm takes a dystonic posture with flexion at the elbow, wrist, and metacarpophalangeal joints. She attempts to suppress the dyskinesia by putting her left arm on her back. She also exhibits backward and right-side tilting of the trunk with retrocollis.

Segment 2. At the follow-up videotape recording performed at the age of 26 years, this patient exhibits marked spontaneous improvement of both the myoclonus and dystonia.

Conflicts of Interest

The authors have no financial conflicts of interest.

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