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Tremor and Other Hyperkinetic Movements

Case Report

Gait Impairment in Myoclonus–Dystonia (DYT-*SGCE*)

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Abstract

Background: Myoclonus–dystonia usually presents variable combination of myoclonus and dystonia mainly affecting the neck and arms, but leg involvement, especially as the presenting sign, is not common.

Case report: A 29-year-old lady with a heterozygous mutation in Epsilon-sarcoglycan (*SGCE*) gene is presented with rapid jerks of the right leg interfering with walking. She has also manifested dystonic posture and jerks of the trunk and proximal upper limbs.

Discussion: Although it is not typical, leg involvement could be a manifestation of myoclonus–dystonia either at presentation or during disease progression.

Keywords: Gait, Epsilon-sarcoglycan, myoclonus–dystonia, myoclonus, DYT11

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Ethics Statement: All patients that appear on video have provided written informed consent; authorization for the videotaping and for publication of the videotape was provided.

Background

Myoclonus–dystonia syndrome (MDS) is a relatively rare syndrome characterized by the combination of myoclonus and dystonia, which typically presents in childhood.[1] There is a broad spectrum of clinical presentations even within a family.[2] It is mainly characterized by upper trunk myoclonic jerks predominantly in proximal muscles along with milder dystonia involving cervical or brachial regions.[3–5]

The most prominent and disabling clinical presentation is myoclonus.[6]

The leading causative gene is Epsilon-sarcoglycan (*SGCE*) on chromosome 7q21–q31, which accounts for 40% of the MDS cases with autosomal dominant inheritance.[1,7] Despite the variations of clinical phenotypes in this syndrome,[4,5] leg involvement as the presenting symptom is unusual.[3–5]

Herein we report a case of MDS with severe leg involvement as the initial and most disabling symptom.

Case presentation

A 29-year-old woman came to our clinic for further assessment of her symptoms. Her problem started when she was 3.5-year-old with involuntary jerky movements in her right leg mostly during walking and running. These movements gradually progressed and affected left leg, upper limbs, trunk, and neck. There was mild improvement of the symptoms with Trihexyphenidyl, Clonazepam, and Tetrabenazine. She also found dramatic improvement of the symptoms by taking alcohol. Her parents were cousins and her brother and paternal uncle had experienced similar symptoms.

Video **1. Rapid Jerks (Myoclonus) of the Right Leg.** She also has dystonic posture of left leg and right hand. Dystonic posture in trunk (causing retropulsion) makes her gait impairment more severe.

On examination she was cognitively intact and speech was normal. She had prominent myoclonic jerks in the right leg that caused imbalance and impending to fall episodes when she was standing or walking. She also had less severe jerks in the neck, trunk, and the arms with dystonic postures in trunk (causing retropulsion) and gait impairment (Video 1). The rest of neurologic examination revealed no abnormality.

Brain MRI was normal. Genetic study revealed heterozygous mutation of *SGCE* gene [C.289C>T (P.R, 77)] confirming diagnosis of myoclonus–dystonia (DYT-*SGCE*).

Discussion

MDS is a combination of dystonia and myoclonus. The disease onset is usually in the first or second decade of life.[3,6]

Action myoclonus is typically present at disease onset and the most disabling symptom, whereas dystonia is the first or associated symptom in 20 and 50% of the patients, respectively.[3] Dystonia is mainly cervical or brachial and interestingly is not the patients' main complaint and mostly it has been detected only on examination.[6] Myoclonus, not dystonia, responds dramatically to alcohol.[8,9] The disease courses in *SGCE*-mutated patients might be benign and not interfere with a normal active life,[10] but there are many exceptions even within the same family.[9,11]

In most reported families, the worsening of myoclonus is the indicator of disease progression,[6] whereas dystonia usually remains stable during patient's life.[1] There are few reports indicating sexual differences in MDS. Affected females may have an earlier onset and more probability of leg involvement in contrast to more common involvement of upper limbs and trunk in males.[1,2]

The involvement of lower limbs at disease onset is unusual, but had been reported in the literature.[3,4,12] In the largest cohort study on MDS patients, of 27 patients with *SGCE* gene mutation, one case (5%) had leg myoclonus and eight cases (42%) had leg dystonia at disease onset, all of them presented before 10 years of age that is similar to our patient. Based on this study, Peal et al. categorized the MDS patients into three main subgroups: the first group, the most common form with predominantly upper body involvement starting before the age of 10 years; the second group, less common form, with predominant lower limb dystonia presenting before the age of 10 years; and the final group, with more prominent cervical dystonia, developing after the age of 10 years.[4]

Raymond et al. in 2008 studied 11 families with MDS and reported six out of seven leg-dominant cases were female and the first symptom was dystonia except for one case who presented with early-onset myoclonus.[3]

Koukouni et al. reported two sisters with abnormal gait and difficulty in standing or walking started at the age of 18 months with progression to upper limbs and neck.[2]

Some authors suggested age as a determining factor in disease topography, in other words the earlier disease onset, the more probability of lower limb involvement and gait impairment.[13,14]

Roze et al. studied 41 patients and found 9 patients had lower limb dystonia and only in 2 patients it was the most prominent affected site.[5]

In another study, among 38 *SGCE* mutation carriers, Tezenas et al. reported lower limb dystonia and myoclonus in five and eight cases, respectively.[12]

Overall, it is thought that more frequent leg involvement has been seen in *SGCE* mutation carriers in contrast to nonmutation carriers.[12]

In conclusion, clinicians should be aware of the possibility that, although not typical, leg involvement can be a manifestation of DYT-*SGCE* either at presentation or during disease course and contribute to a significant gait impairment.

Author contribution

Ghazal Haeri was responsible for rimary drafting of the article. Gholamali Shahidi did critical revision of the manuscript. Alfonso Fasano was responsible for the interpretation and critical revision of the manuscript for important intellectual content. Mohammad Rohani was responsible for the study concept and design, acquisition of data, and primary drafting.

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