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Tremor and Other Hyperkinetic Movements

Brief Report

Adult-onset Idiopathic Focal Lower Extremity Dystonia: A Rare Task-Specific Dystonia

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Abstract

Background: Adult-onset focal lower extremity (LE) dystonia is rare, but there have recently been a number of case series that have reported an idiopathic variant triggered during ambulation.

Methods: We describe nine patients with idiopathic, focal task-specific LE dystonia. We conducted a comparative analysis that included our cohort and several recently published case series to further characterize the disorder.

Results: A total of 48 patients (37 female, 11 male) were compared. The average age of onset was 48 years; 36 patients had distal extremity involvement (75%), 5 proximal (10%), and 7 both proximal and distal (15%). Among 33 patients in which the dystonic side was known, 20 were affected on the left (61%). Inversion of the foot with flexion of one or more toes was the most prevalent pattern in those with distal extremity involvement.

Discussion: This is a novel task-specific dystonia triggered during ambulation that is often misdiagnosed as an orthopedic or psychogenic issue.

Keywords: dystonia, task specific, focal, lower extremity

Citation: Ramdhani RA, Frucht SJ. Adult-onset idiopathic focal lower extremity dystonia: a rare task-specific dystonia. Tremor Other Hyperkinet Mov 2012;2: http://tremorjournal.org/article/view/142

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Editor: Elan D. Louis, Columbia University, United States of America

Received: October 23, 2012 **Accepted:** November 26, 2012 **Published:** January 22, 2013

Funding: None.

Financial Disclosures: None.

Conflict of Interest: The authors report no conflicts of interest.

Introduction

Focal task-specific dystonias (FTSDs) usually affect the upper limbs or craniocervical regions (e.g., writer's cramp, musician's hand dystonia, or embouchure dystonia). Lower extremity (LE) dystonia is thought to be rare in adults and more common in children. When it does occur in adults, it tends to be non-task specific and is often associated with parkinsonism, trauma, stroke, or psychogenic behavior. Over the last several years, numerous case reports[1–3] and series[4–7] have highlighted an unusual task-specific dystonia with ambulatory triggers. Here, we describe nine patients with idiopathic, focal task-specific LE dystonia and compare them to several recently published case series to call attention to the condition.

Case series

Among the patients we encountered (Table 1), the average age was 49.3 years with females outnumbering males (2:1). The average age of symptom onset was 45.6 years with an average time to diagnosis of 3.7 years. Many patients had extensive workups, including normal imaging studies, prior to being seen in our center. None had received dopamine receptor blocking agents or had experienced prior central nervous system insult.

Lower limb dystonia (five right, four left) was triggered by ambulation in all nine patients and was not present at rest. Six patients had distal extremity involvement, two patients demonstrated proximal involvement (i.e., hip flexion, hyperextension, and abduction), and one patient exhibited both proximal and distal extremity involvement (i.e., hip flexion with foot plantar flexion). Foot inversion was the most common pattern (4/9 patients), with two patients having concomitant plantar flexion. One patient (Patient 9) had involvement of the toes along with foot inversion. Walking backward or simply imagining walking backward while walking forward often improved symptoms. One patient (Patient 7) had coincident R wrist 'yips' or

Table 1. Mount Sinai Case Series

Patient #	Age/ Sex	Onset Age	Duration (yrs)	Limb	Precipitant Movement Task	Phenomenology	Present at Rest	Other Limbs Affected	FH	Medications	Response
1	31/M	26	5	R	Walking down steps	Plantarflexes foot and flexes hip when walking down steps. Absent while walking up steps or backward.	No	None	PD	Sinemet	None
2	24/F	20	4	L	Marathon running	Inversion and plantarflexion of the foot while walking that becomes more prominent with running. Does not occur when walking backward.	No	None	None	Sinemet	None
3	33/F	31	2	R	Marathon running	Sustained inversion while walking, which normalizes while walking backward. Improves with side stepping in either direction as well as walking forward while imagining walking backward.	No	None	None	Sinemet Artane Baclofen BTX to calf Valium	None None None None Yes
4	60/F	58	2	L	Distance walking	Hip flexion with slight side to side lurch while walking. Improves with imagined walking backward while walking forward.	No	None	Mother & Father - PD	Klonopin Artane	None None
5	61/M	58	3	R	Walking	Eversion of foot while walking on cobblestone terrain or walking down steps. Disappears while walking on flat surfaces, running, or climbing stairs.	No	None	Mother - similar movements	Sinemet	None
6	27/F	22	5	L	Distance running	Inversion and sustained plantarflexion of the foot while walking. Movements disappeared while walking backward ,as well as imagined walking backward while walking forward. No abnormal movements when climbing stairs or during side stepping.	No	None	None	Artane	None

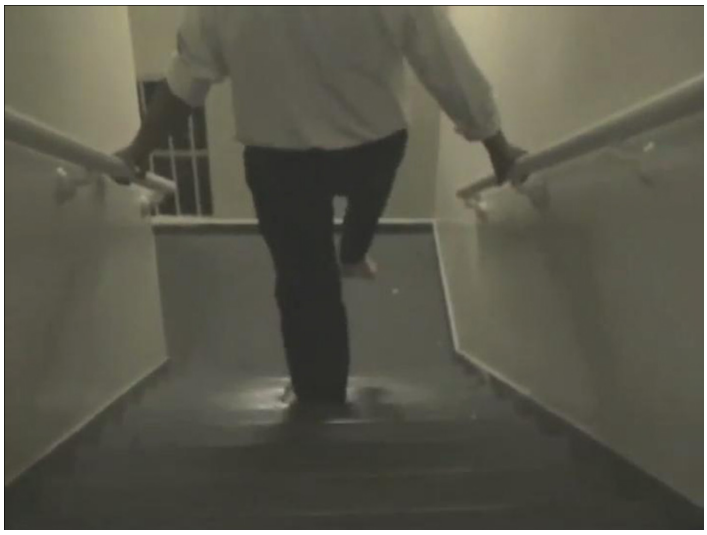
Table 1. Continued

Patient #	Age/ Sex	Onset Age	Duration (yrs)	Limb	Precipitant Movement Task	Phenomenology	Present at Rest	Other Limbs Affected	FH	Medications	Response
7	80/M	78	2	R	Walking	Eversion of foot while walking that disappears while walking backward or sideways and climbing stairs.	No	Yes - R Wrist 'Yips'	None	Sinemet	None
8	53/F	49	4	L	Walking	Curling of toes and inversion of foot while walking, which is also produced with foot tapping.	No	None	None	BTX Klonopin Gabapentin	Yes Yes Yes
9	75/F	69	6	R	Walking	Hyperextension and abduction of leg. More prominent with speed walking.	No	None	None	Sinemet Ropinerole	None None
Mean	49.3	45.6	3.7								

Table 2. Focal Task-Specific Lower Extremity Dystonia Comparative Analysis

Case Series	Patients	Sex		Idiopathic Cases		Average Age of Onset	FH of Movement Disorders	Limb Affected (Idiopathic)			Other Body Parts	Present at Rest
	No.	M	F	M	F	(yrs)		R	L	Both		
Singer and Papapetropoulus[4]	4	1	3	1	3	59.5	x	0	4	0	0/4	2
Schneider et al.[5]	17	6	11	5	11	48.2	PD - 1 LD -2 Hand Tremor - 1	7	8	1	2/15 (arm, cervical dystonia)	NR
Wu and Jankovic[6]	5	2	3	2	3	37.4	0	1	4	0	0/5	0
McKeon et al.[7]	36	5	31	0	14	51.1	NR	NR	NR	NR	0/14	4
MSMC	9	3	6	3	6	45.7	PD - 2 LD - 1	5	4	0	1/9(yips)	0
Total	71	17	54	11	**37**	**48.38**		**13**	**20**	**1**		**6**

Abbreviations: (FH, Family History; PF, Plantarflexion; DF, Dorsiflexion; (+), presence; (−), absence; NR, not reported; PD, Parkinson[1]s Disease; LD, Limb dystonia; BTX, Botulinum Toxin; MSMC, Mount Sinai Medical Centre).



Video. Patient 1 has Right Hip Flexion while Walking Down Steps.

Video. Patient 3 has Inversion and Sustained Plantar Flexion of the Left Foot while Walking Forward.

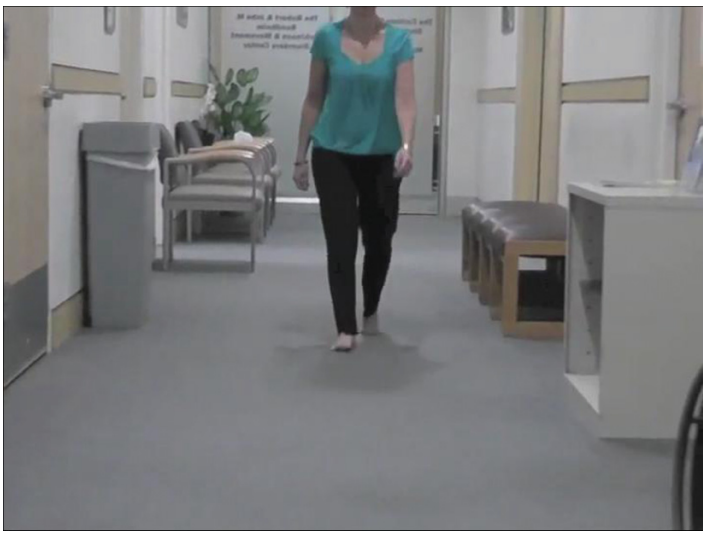
golfer's cramp. Treatment with L-dopa (0/5 patients) was ineffective, but one of two patients treated with botulinum toxin injections improved.

Adding our cases to those published previously yielded a total of 48 patients (37 female, 11 male) diagnosed with idiopathic FTSD of the LE. Comparative analysis of these patients (Table 2) revealed that 36 had distal extremity involvement (75%), 5 demonstrated proximal (10%), and 7 both proximal and distal (15%). The average age of symptom onset was 48 years old, and family history of dystonia was rare. Among 33 patients in whom the side of the dystonia was known, 20 were affected on the left (61%). Routine ambulatory tasks (e.g., standing, walking, ascending or descending stairs) were triggers of the dystonic limb posture, and only six patients (13%) had dystonia at rest.

Concomitant upper extremity dystonia was observed in three patients, and one had cervical dystonia.

Analysis of the 43 patients with distal extremity involvement revealed that 29 (67%) had a predilection for foot inversion, and 14 of those 29 patients (48%) exhibited concomitant plantar flexion. Pure ankle eversion (three patients), foot plantar flexion (four patients) and foot dorsiflexion (two patients) were less common. Toe movements described as either dorsi- or plantar flexion of one or more toes were observed in 21 of 43 patients (49%), with 19 of those 21 patients (90%) having associated dystonic posturing of the foot.

Video. Patient 2 Exhibits Right Foot Inversion while Walking Forward.



Video. Patient 4 has Inversion of the Left Foot with Toe Curling while Walking Forward.

Compared to other types of FTSD, LE dystonia manifests at a later age and predominantly affects females (F): Musician's hand (36 yrs/F20%),[8] Writer's Cramp (38 yrs/F33%),[8] Embouchure (36 yrs/F28%),[8] Cranial (46 yrs/F43%),[8] LE (48 yrs/F77%). These FTSDs share features such as lack of spread to other limbs and absence of trauma as an underlying factor.

Discussion

All nine patients in our case series had unremarkable medical histories; none exhibited signs of psychogenic behavior,[9,10] such as fixed dystonia, inconsistency, incongruency, or weakness upon examination. There were no histories of mood disorders or pending litigation. Parkinsonism was not present at onset and had not developed during 4 years of follow up. This is similar to what was observed in the other four cases series. Because DYT-1 dystonia[11] usually begins in the LE, especially in those younger than 26 years, Patients 1 and 6 were tested and found to be negative, whereas Patients 2 and 3 were referred for testing but did not do so.

LE movements had several factors in common in all 48 patients of the comparative analysis. First, various ambulatory tasks (e.g., walking, walking down steps, running) were specific triggers for the dystonic posturing, which was rarely present at rest and failed to spread to other limbs. Second, exteroceptive sensory gestes, such as walking backward and side stepping, as well as the interoceptive sensory geste[12] of imagining an external sensory trick (e.g., walking backward) while simultaneously walking forward led to improvement in 6 of 9 patients in our series, as well as many others in the cumulative series (2/4 patients in Singer and Papapetropoulus,[4] 3/5 patients in Wu and Jankovi,[6] and 6/14 patients in McKeon et al.[7]).

The pathophysiology of this dystonia is unclear, and like most other dystonias, it is the result by both genetic and environmental factors.[13] In 15 patients, the initial presentation occurred during prolonged exercise, long distance running, or long distance walking, suggesting a transformation from intense exertion-induced dystonia to one that is easily triggered by routine ambulation. Although it can also be argued that this is a phenomenon of paroxysmal kinesogenic or exertion-induced dyskinesias, the constancy and immediate triggering of the dystonia suggests otherwise. The later age of onset, as well as the considerable number of patients who developed dystonia in the setting of prolonged exertion, further insinuates that repetitive use of a limb[14] may peripherally stimulate a change in cortical and/or striatal plasticity, perhaps triggering the dystonic network. The loss of specificity over time may also be a manifestation of surround inhibition failure.[13]

It is important that physicians recognize this entity as a task-specific dystonia triggered by various modes of ambulation. Many of our patients were misdiagnosed as having orthopedic problems or manifesting psychogenic behavior. The later age of onset, female predominance, and specific foot movement patterns of plantar flexion, inversion, and toe(s) involvement with sensory geste reduction offer clues to its diagnosis. Botulinum toxin injections seem to be most effective treatment across all case series.

References

1. Garcia-Ruiz PJ, del Val J, Losada M, et al. Task-specific dystonia of the lower limb in a flamenco dancer. *Parkinsonism Relat Disord* 2011;17:221–222, http://dx.doi.org/10.1016/j.parkreldis.2010.12.001.

2. Duarte J, Sempere AP, Coria F, et al. Isolated idiopathic adult-onset foot dystonia and treatment with botulinum toxin. *J Neurol* 1995;242:114–115, http://dx.doi.org/10.1007/BF00887828.

3. Shimizu T, Shiraishi M, Hirayama T, et al. [A case of a middle-age man with equinovarus movement at the end of the swing phase of walking]. *Rinsho Shinkeigaku* 2002;42:895–897.

4. Singer C, Papapetropoulos S. Adult-onset primary focal foot dystonia. *Parkinsonism Relat Disord* 2006;12:57–60, http://dx.doi.org/10.1016/j.parkreldis.2005.07.004.

5. Schneider SA, Edwards MJ, Grill SE, et al. Adult-onset primary lower limb dystonia. *Mov Disord* 2006;21:767–771, http://dx.doi.org/10.1002/mds.20794.

6. Wu LJ, Jankovic J. Runner's dystonia. *J Neurol Sci* 2006;251:73–76, http://dx.doi.org/10.1016/j.jns.2006.09.003.

7. McKeon A, Matsumoto JY, Bower JH, et al. The spectrum of disorders presenting as adult-onset focal lower extremity dystonia. *Parkinsonism Relat Disord* 2008;14:613–619, http://dx.doi.org/10.1016/j.parkreldis.2008.01.012.

8. Frucht SJ. Embouchure dystonia--Portrait of a task-specific cranial dystonia. *Mov Disord* 2009;24:1752–1762, http://dx.doi.org/10.1002/mds.22550.

9. Fahn S, Williams DT. Psychogenic dystonia. *Adv Neurol* 1988;50:431–455.

10. Schrag A, Trimble M, Quinn N, et al. The syndrome of fixed dystonia: an evaluation of 103 patients. *Brain* 2004;127:2360–2372, http://dx.doi.org/10.1093/brain/awh262.

11. Bressman SB, Sabatti C, Raymond D, et al. The DYT1 phenotype and guidelines for diagnostic testing. *Neurology* 2000;54:1746–1752, http://dx.doi.org/10.1212/WNL.54.9.1746.

12. Greene PE, Bressman S. Exteroceptive and interoceptive stimuli in dystonia. *Mov Disord* 1998;13:549–551, http://dx.doi.org/10.1002/mds.870130329.

13. Hallett M. Neurophysiology of dystonia: The role of inhibition. *Neurobiol Dis* 2011;42:177–184, http://dx.doi.org/10.1016/j.nbd.2010.08.025.

14. Frucht SJ. Focal task-specific dystonia in musicians. *Adv Neurol* 2004;94:225–230.