Original Article

Spectrum of Truncal Dystonia and Response to Treatment: A Retrospective Analysis

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Abstract

Background: Presence of truncal dystonia usually points to a secondary cause of dystonia like exposure to dopamine receptor blockers or neurodegenerative illness. Rarely, it can occur as an idiopathic focal or segmental dystonia. **Methods:** Retrospective review of medical records and videos of patients of truncal dystonia presenting in the Botulinum Toxin Clinic of Department of Neurology at Post Graduate Institute of Medical Education and Research, Chandigarh between May 2016 and February 2019. **Results:** A total of 16 patients with predominant truncal dystonia were recruited. There were ten males and six females with mean age of 49.1 ± 15.1 years (range 22–70). Extensor truncal dystonia was the most common (12/16) followed by camptocormia (4/16). Various etiologies included Idiopathic Parkinson's disease (4/16), Tardive dystonia (5/16), Neurodegeneration with brain iron accumulation (genetically confirmed) (2/16) and idiopathic (5/16). All patients were refractory to a combination of oral medications tried over a period of 1.82 ± 1.93 years. All patients received electromyographic-guided botulinum toxin in paraspinals or rectus abdominis muscles depending upon the type of dystonia. The mean dose of abobotulinum toxin used was 286.7 ± 108.6 units (range 200–500 units) for paraspinals and 297.5 ± 68.5 (range 200–350) for rectus abdominis muscles per session. Average subjective response after botulinum toxin injection session was 31.2 ± 21.5% (range 0–70). No adverse effects were reported. **Conclusion:** Botulinum toxin is an acceptable alternative to patients presenting with medically refractory truncal dystonia and may offer modest benefit.

Keywords: Botulinum toxin, camptocormia, Parkinson's disease, Truncal dystonia

Introduction

Truncal dystonia is characterized by involuntary contractions and postures of the paraspinal, abdominal, and chest muscles.[1] Presence of truncal dystonia usually points to a secondary cause of dystonia-like exposure to dopamine receptor blockers or several neurodegenerative conditions.[2] Rarely, it can also present as an idiopathic focal or segmental dystonia.[3] Depending on the direction of the dystonic movement, it can be classified into flexion, extension, or lateral flexion types. Camptocormia is defined as forward flexion of the thoracolumbar spine (>45 degrees) which is characterized by overactivation of the rectus abdominis muscles.[4] Opisthotonus is defined as an extension posturing of the spine resulting in arching of the back due to overactivation of thoracic and lumbar paraspinal/paravertebral muscles.[1] Lateral flexion of the spine also known as the Pisa syndrome occurs due to overactivity in the internal and external oblique muscles in conjunction with anterior abdominal and paravertebral muscles.[5]

Truncal dystonia usually responds poorly to medical treatment in the form of dopaminergic therapy, anticholinergics and various other drugs used in the armamentarium to treat dystonia.[1] It can seriously impair the quality of life causing difficulty in walking, pain and if untreated, can lead to fixed postures and deformities. Physical therapy, botulinum toxin treatment, and deep brain stimulation have each been tried in small number of cases with variable results.[6-8]

Botulinum toxin treatment of the dystonic muscles have shown to improve the abnormal posture, pain associated with abnormal contractions and gait with persistent improvement at 3 months.[1,6,9,10] However, the presentation of dystonia and pattern of improvement in each of the published studies was different from each other.[1,6,9,10] With this background in mind, we attempted to identify the clinical and demographic profile of patients presenting with truncal dystonia, characterize the pattern of involvement and assess the response to treatment from a tertiary care hospital.

Methods

We retrospectively reviewed the medical records and videos of more than 600 patients who presented to the Botulinum toxin

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clinic of Department of Neurology at Post Graduate Institute of Medical Education and Research, Chandigarh between May 2016 to February 2019. Patients with predominant truncal dystonia were included in the study. Truncal dystonia could exist in isolation or in association with dystonia in other body parts. Patients who did not achieve substantial clinical benefit from a trial of oral medications for at least 3 months were selected to be treated with botulinum toxin. Abobotulinum toxin was used in all the patients and the oral medications were continued throughout the treatment period. Abobotulinum toxin was reconstituted with 2.5 ml of unpreserved normal saline (concentration of 20 U per 0.1 ml). Posterior paraspinal muscles were selected for extensor truncal dystonia and rectus abdominis muscles were selected for camptocormia. Both the paraspinals and rectus abdominis muscles were injected under electromyographic guidance. The number of injection sites and dose was individualized depending upon the clinical severity and/or degree of muscle hypertrophy. All the patients were reassessed and videotaped 1 month after the injections when the maximal benefit of botulinum toxin is expected to occur. The outcome was assessed based on a single question evaluating improvement in pain, dystonia and functional status and was scored between 0 and 100. Improvement was assessed only for truncal dystonia in patients with multifocal, segmental, or generalized distribution. The response was measured only for the first injection session in patients who underwent multiple sessions. The study was approved by institute ethics committee (INT/IEC/2020/SPL-419). Patients and their caregivers signed informed consent for botulinum toxin injections.

Results

A total of sixteen patients with predominant truncal dystonia were found. There were ten males and six females. Mean age was 49.1 ± 15.1 years (range 22–70 years). Various etiologies included Idiopathic Parkinson's disease (4), Tardive (5), Idiopathic (5), and Pantothenate kinase associated neurodegeneration (2) [Table 1]. Mean duration of the disease was 4.63 ± 3.7 years (range 2 months–14 years). The mean interval between the onset of parkinsonism and truncal dystonia was 17 ± 9.5 months (4–24 months) in patients with IPD. Ten patients exhibited dystonia in other body parts in addition to truncal dystonia. Three patients had blepharospasm, ten patients had associated cervical dystonia and eight patients had associated involvement of pectoral muscles. Extensor truncal dystonia was more commonly found than flexion predominant dystonia [Table 1]. Patients were tried on multiple medications in combination before proceeding to botulinum toxin injections. These included trihexiphenidyl (9 patients), tetrabenazine (7 patients), clonazepam (11 patients), baclofen (3 patients), levodopa (8 patients), and carbamazepine (1 patient). The mean duration of medications before the first session of botulinum toxin injection was 1.82 ± 1.93 years (range 2 months–6 years). The mean number of medications was 2.8 ± 0.6 (range 1–6). The mean improvement with medications was 13.8 ± 12.7% (range 0–30) [Table 1]. No relief or suboptimal relief with oral medications was the main indication for botulinum toxin injections. The mean dose of abobotulinum toxin used was 286.7 ± 108.6 units (range 200–500 units) for paraspinals and 297.5 ± 68.5 (range 200–350) for rectus abdominis muscles per session. Average subjective response after botulinum toxin injection session was 31.2 ± 21.5% (range 0–70) [Videos 1 and 2]. One patient of Idiopathic Parkinson's disease with camptocormia and another with idiopathic dystonia did not show any subjective improvement in their dystonia. The mean duration of effect was 3 months. The mean number of botulinum toxin injection sessions were 2.5 ± 1.6 (range 1-7). The mean interval between the injections was 4.8 ± 1.9 months (range 3–9) [Table 1]. The mean dose of abobotulinum toxin injected in the different muscles is described in Table 1. No adverse effects were reported in our patients. The response with botulinum toxin injection was almost the same in all types of dystonia and for all pathological indications averaging about 30–35% [Table 1].

Discussion

Truncal dystonia can occur in isolation or in combination with dystonia in other body parts in a segmental, multifocal or generalized pattern.[3] It is much more commonly seen as a part of neurodegenerative illness like Parkinson's disease, atypical parkinsonian syndromes, neurodegeneration with brain iron accumulation or as a tardive syndrome secondary to antipsychotics. However, it can also occur as an idiopathic variety as reported previously.[3,11] The largest case series of eighteen patients of axial predominant primary truncal dystonia is reported by Bhatia *et al*.[3] Ehlrich *et al*. reported seven patients of idiopathic truncal dystonia.[11] In both the series, flexion was the most common direction of dystonic movement. Around one-third of patients in our series belonged to the idiopathic variety. Truncal extension was the most common direction of dystonic movement in contrast to the reports in the literature where flexion is more commonly reported.[3,11]

Around one-third of patients with Parkinson's disease can manifest dystonia.[12] It is more prevalent in young onset Parkinson's disease and can occur in untreated PD patients or as a complication of dopaminergic therapy. Camptocormia and Pisa syndrome can occur in both PD and atypical parkinsonism particularly multiple system atrophy.[5] As reported by Bonanni *et al*., prevalence of axial dystonia was 4.4% in their cohort of 1400 patients of Parkinsonism. Out of these, camptocormia was identified in 2.6% and lateral axial dystonia in 1.9% in patients presenting with dopa responsive Parkinsonism in their study.[9] Four patients in our series were suffering from Parkinson's disease; 2 had camptocormia and 2 had extensor truncal dystonia.

Five patients developed truncal dystonia as a result of exposure to dopamine receptor blockers. The drugs included trifluoperazine, haloperidol, risperidone, and levosulpiride. Extensor truncal dystonia was much more common which is consistent with the existing literature.[13] Only one patient developed levosulpiride-induced camptocormia. She had normal 18F DOPA

Table 1: Patient Characteristics

Total number of patients (*n*)	16		
Male: Female	10:6		
Mean age±SD (Range)	49.1±15.1 (22-48)		
Mean duration of illness (In years)	4.63±3.71 (0.2- 14)		
Primary diagnosis (*n*)	**Flexion**	**Extension**	**Total**
IPD	2	2	4
Tardive	1	4	5
Idiopathic	1	4	5
PKAN	0	2	2
Flexion: Extension	4:12		
Botulinum Toxin details Mean±SD, Range			
Improvement before abobotulinum toxin (in %)	13.9±12.7 (0- 30)		
Duration of medications before abobotulinum toxin (In years)	1.8±1.9 (0.2 – 6)		
Number of doses	2.5±1.6 (1-7)		
Average dose interval (In months)	4.8±1.9 (3-9)		
Average dose of abobotulinum toxin	289.4±98.1 (200- 500)		
Onset of effect (days)	8.8±6.2 (0- 20)		
Peak effect (Months)	1.6±1.6 (0-6)		
Total duration of effect (months)	2.9±3.1 (0-12)		
Effect (%)			
Total	31.2±21.5 (0- 70)		
Flexion	32.5±23.6 (0-50)		
Extension	30.8±21.8 (0-70)		
Amount of Abobotulinum toxin injected in different muscles Mean±SD, Range	**Unilateral**	**Bilateral**	
Paraspinals (*n*=12)	143.33±54.32 (100-250)	286.67±108.65 (200-500)	
Rectus abdominis (*n*=4)	148.75±34.24 (100-175)	297.50±68.49 (200- 350)	
Mean Subjective improvement in % with Abobotulinum toxin according to etiology (No of subjects, *n*)			
IPD (4)	35%		
Tardive (5)	32%		
Idiopathic (5)	29%		
PKAN (2)	25%		

SD: Standard deviation; IPD: Idiopathic Parkinson's disease; PKAN: Pantothenate Kinase Associated Neurodegeneration

PET scan thus essentially ruling out parkinsonism.

Two of our patients had genetically proven neurodegeneration with brain iron accumulation (PANK2 mutation positive). Both these patients presented with action induced dystonic opisthotonus in their thirties. MRI brain showed the characteristic "eye of the tiger sign". Dystonic opisthotonus can be a clinical clue to the diagnosis of neurodegeneration with brain iron accumulation.[14]

Truncal dystonia is highly disabling and is usually refractory to combination of multiple oral medications. Our patients showed only subjective improvement of 13% with a combination of oral drugs.

Comella *et al*. first reported the use of botulinum toxin for extensor truncal dystonia.[1] They reported mean improvement of 37% 1 month after the botulinum toxin injection. Maximum improvement occurred in the pain secondary to dystonia to the extent of 65%. Patients in our series also reported a modest benefit of 31%. Bonnani *et al*. studied the efficacy of botulinum toxin in lateral axial dystonia in levodopa responsive parkinsonism in their double-blind randomized cross over study.[9] Patients receiving botulinum toxin showed an improvement of 50–85.7% while there was no improvement in placebo group. Other case series on efficacy of botulinum toxin in truncal dystonia are tabulated in Table 2.

Main limitation of our study is that it was a retrospective analysis. Second, the response to treatment was subjective based on patient responses and not objective based on video and scales.

Conclusion

Truncal dystonia is a relatively rare presentation. Although it commonly presents as secondary dystonias, it may occur as an idiopathic variety. Both flexion and extension varieties can be seen. Botulinum toxin may be used as an effective and safe option in the treatment of this condition that is usually refractory to oral medications.

Statement of informed consent

The patients signed informed consent for publication in a medical journal.

Table 2: Synopsis of reported case series and case reports on use of botulinum toxin in truncal dystonia

Case Series/ Report, Year	No. of Cases	Type of truncal dystonia	Muscle Injected	Diagnosis	Dose and type of botulinum toxin	Response to Botulinum toxin
Comella *et al*., 1998[1]	5	Extensor	Paravertebral muscles	Dystonia	150-300 units (Onabotulinum toxin type A)	Objective improvement on videotape scores (37%); subjective improvement (46%); 30-80% improvement in pain
Azher *et al*., 2005[4]	16	Camptocormia	Rectus abdominis and / or Paraspinals	PD (11) 5 other MD	(9 patients injected) PD group (6):350-600 units Onabotulinum toxin A Non PD group (3): 300-800 unit Onabotulinum toxin A	PD group: Good response- 3, No response- 3 Non PD group: Partial response- 1 No response-2
Tassorelli *et al*., 2014[6]	26	Pisa Syndrome (Lateral trunk deviation) plus camptocormia	Iliopsoas, Rectus abdominis, Paravertebrals	Parkinson's disease	50-200 units (Incobotulinum toxin type A)	Lateral trunk inclination significantly reduced in the Botulinum toxin group (13 patients) versus saline group (13 patients)
Bonanni *et al*., 2007[9]	9	Lateral Axial Dystonia	Paraspinal muscles	Parkinson's disease	500 Units (Abobotulinum toxin type A)	Improvement in 6 out of 9 patients Improvement of posture by 50-85.7% as measured by goniometric scale; improvement in pain (24.1-76.4%)
Todo *et al*., 2018[10]	6	Camptocormia	External oblique muscle	Parkinson's disease	150-180 units (Onabotulinum toxin type A)	Significant attenuation of camptocormia angle (38 degrees versus 18 degrees at 2 weeks)
Fietzek *et al*., 2009[15]	10	Camptocormia	Iliospoas and Rectus abdominis (B/L)	PD	100-300-unit Incobotulinum toxin A	No improvement in goal attainment scales incorporating pain relief, postural improvement, functional goals at 3 weeks.
Colosimo *et al*., 2009[16]	2	Camptocormia	Iliopsoas (B/L) B/L RA CT guided	PD	800 units Onabotulinum toxin A	No response over 2 weeks
Von Coelln *et al*., 2008[17]	4	Camptocormia	Iliopsoas (UL 2, B/L 2) (USG guided)	PD-3 MSA-1	1000-3000 Units Abobotulinum toxin A	Improvement in posture after 2 weeks —1 (NS) Improvement in posture after 6 weeks -1(NS) No improvement- 2
Wijemanne *et al*., 2014[18]	1	Camptocormia	Rectus abdominis (B/L) Unilateral RA Contralateral EO	PD	400 units Onabotulinum toxin A	RA- improved pain, minimal posture change. RA + EO: camptocormia improved from 45° to 15°-20° forward flexion in the 'on' motor state.
Yadav *et al*., 2015[19]	1	Camptocormia	Rectus abdominis and/or Iliopsoas (B/L)	Idiopathic dystonic camptocormia	200 units botulinum toxin A	Effective only for transient period
Mehta *et al*. (This study)	16	Camptocormia and Opisthotonus	Rectus abdominis and Paraspinals	PD-4 Tardive- 5 Idiopathic-5 PKAN-2	200- 500 units (Abobotulinum toxin type A)	Mild to moderate subjective improvement

PD: Parkinson's disease; MD: Movement Disorders; CT: Computed Tomography; B/L: Bilateral; MSA: Multiple System Atrophy; PKAN: Pantothenate Kinase Associated Neurodegeneration; RA: Rectus abdominis; EO: External oblique; UL: Unilateral; NS: not significant

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Conflicts of interest

There are no conflicts of interest.

References

1. Comella CL, Shannon KM, Jaglin J. Extensor truncal dystonia: Successful treatment with botulinum toxin injections. Mov Disord 1998;13:552-5.
2. Stamelou M, Lai SC, Aggarwal A, Schneider SA, Houlden H, Yeh TH,

et al. Dystonic opisthotonus: A 'red flag' for neurodegeneration with brain iron accumulation syndromes? Mov Disord 2013;28:1325-9.
3. Bhatia KP, Quinn NP, Marsden CD. Clinical features and natural history of axial predominant adult onset primary dystonia. J Neurol Neurosurg Psychiatry 1997;63:788-91.
4. Azher SN, Jankovic J. Camptocormia: Pathogenesis, classification, and response to therapy. Neurology 2005;65:355-9.
5. Barone P, Santangelo G, Amboni M, Pellecchia MT, Vitale C. Pisa syndrome in Parkinson's disease and parkinsonism: Clinical features, pathophysiology, and treatment. Lancet Neurol 2016;15:1063-74.
6. Tassorelli C, De Icco R, Alfonsi E, Bartolo M, Serrao M, Avenali M, *et al*. Botulinum toxin type A potentiates the effect of neuromotor rehabilitation of Pisa syndrome in Parkinson disease: A placebo controlled study. Park Relat Disord 2014;20:1140-4.
7. Bartolo M, Serrao M, Tassorelli C, Don R, Ranavolo A, Draicchio F, *et al*. Four-week trunk-specific rehabilitation treatment improves lateral trunk flexion in Parkinson's disease. Mov Disord 2010;25:325-31.
8. Hagenacker T, Gerwig M, Gasser T, Miller D, Kastrup O, Jokisch D, *et al*. Pallidal deep brain stimulation relieves camptocormia in primary dystonia. J Neurol 2013;260:1833-7.
9. Bonanni L, Thomas A, Varanese S, Scorrano V, Onofrj M. Botulinum toxin treatment of lateral axial dystonia in Parkinsonism. Mov Disord 2007;22:2097-103.
10. Todo H, Yamasaki H, Ogawa G, Nishida K, Futamura N, Funakawa I. Injection of onabotulinum toxin a into the bilateral external oblique muscle attenuated camptocormia: A prospective open-label study in six patients with Parkinson's disease. Neurol Ther 2018;7:365-71.
11. Ehrlich DJ, Frucht SJ. The phenomenology and treatment of idiopathic adult-onset truncal dystonia: A retrospective review. J Clin Mov Disord 2016;3:15.
12. Shetty AS, Bhatia KP, Lang AE. Dystonia and Parkinson's disease: What is the relationship? Neurobiol Dis 2019;132:104462.
13. Krack P, Schneider S, Deuschl G. Geste device in tardive dystonia with retrocollis and opisthotonic posturing. Mov Disord 1998;13:155-7.
14. Mehta S, Lal V. Neurodegeneration with brain iron accumulation: Two additional cases with dystonic opisthotonus. Tremor Other Hyperkinet Mov (N Y) 2019;9:1-3. doi: 10.7916/tohm.v0.683.
15. Fietzek UM, Schroeteler FE, Ceballos-Baumann AO. Goal attainment after treatment of parkinsonian camptocormia with botulinum toxin. Mov Disord 2009;2413:2027-8. doi:10.1002/mds.22676.
16. Colosimo C, Salvatori FM. Injection of the iliopsoas muscle with botulinum toxin in camptocormia. Mov Disord 2009;24:316-7. doi:10.1002/mds.22249.
17. von Coelln R, Raible A, Gasser T, Asmus F. Ultrasound-guided injection of the iliopsoas muscle with botulinum toxin in camptocormia. Mov Disord 2008;23:889-92. doi:10.1002/mds.21967.
18. Wijemanne S, Jimenez-Shahed J. Improvement in dystonic camptocormia following botulinum toxin injection to the external oblique muscle. Parkinsonism Relat Disord 2014;20:1106-7. doi:10.1016/j.parkreldis.2014.06.002.
19. Yadav R, Ansari AZ, Surathi P, Srinivas D, Somanna S, Pal P. Bilateral pallidal deep brain stimulation in idiopathic dystonic camptocormia. Neurol India 2015;63:911-4. doi:10.4103/0028-3886.170080.