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Clinical Spectrum of Focal Dystonia in Patients at Tertiary Care Hospital in Karachi: A Cross-Sectional Study

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Abstract

Objective: To describe the clinical patterns and types of focal dystonia among patients presenting to the neurology department of a tertiary care hospital in Karachi.

Methodology: This descriptive cross-sectional study was undertaken at Dr. Ruth K. M. Pfau Civil Hospital, a tertiary care institution in Karachi, from 21st March 2025 to 20th September 2025, including individuals aged 15 to 60 years of both genders. The exclusion criteria included patients with clinical conditions mimicking dystonia (neck extensor myopathy, post-traumatic deformities, Isaacs syndrome, neuromuscular junction disorders, and hemifacial spasm) and non-consenting patients. Following the acquisition of ethical clearance, participants were selected via convenience sampling methodology, with recruitment of consecutive patients presenting with focal dystonia to our department. Comprehensive data regarding demographic characteristics, clinical manifestations, and classification of focal dystonia (blepharospasm, oromandibular dystonia, laryngeal dystonia, cervical dystonia, and limb dystonia) were systematically gathered. Statistical evaluation was conducted utilizing SPSS software, with a significance threshold established at $p \leq 0.05$, using the Chi-square test.

Results: There were a total of 75 patients, with a mean age of 32.09 ±11.68 years . Most participants were men (59, 78.7%), while 16 (21.3%) were women. The most frequent form of dystonia involved the neck muscles, causing involuntary twisting of the head (cervical dystonia, 28 patients-37.3%). This was followed by dystonia affecting the arms or legs (limb dystonia, 18 -24.0%), eyelid spasms leading to repeated blinking (blepharospasm, 14 patients -18.7%), voice muscle involvement causing a strained or interrupted voice (laryngeal dystonia, 8 patients 10.7%), and jaw or facial muscle contractions (oromandibular dystonia, 7 patients - 9.3%). Dystonia subtype was strongly connected with both gender (p=0.001) and duration (p=0.050) exhibiting different clinical patterns. Concerning the variability of symptoms, 77.3% experienced persistent dystonia, 10.7% had paroxysmal symptoms, and 12.0% reported diurnal fluctuation.

Conclusion: This study identified cervical dystonia as the most frequent form of focal dystonia, followed by limb dystonia and blepharospasm. Beyond defining subtype distribution, these findings highlight the need for improved awareness and earlier recognition of dystonia among general clinicians to prevent diagnostic delays.

Keywords: Focal dystonia; Neurological disorders; Blepharospasm; Cervical dystonia

Introduction

Dystonia is a hyperkinetic movement disorder characterized by involuntary, sustained, and often patterned contractions of muscles, resulting in twisting movements or abnormal postures.[1] These movements are typically repetitive and may be associated with overflow activation of adjacent muscles, which helps differentiate dystonia from other movement disorders.[2] Etiologically, dystonia is classified as primary (idiopathic), where no structural or metabolic cause is evident, or secondary (symptomatic), which may be related to underlying lesions, systemic disease, or drug exposure.[3,4] Anatomically, it can present as focal, segmental, multifocal, or generalized, with focal dystonia being the most frequently encountered in outpatient neurology clinics.[5,6]

Dystonia is termed as "focal dystonia" when it involves only one anatomical portion of the body.[1] The global prevalence of idiopathic focal dystonia ranges from 30 to 732 per 100,000 population, reflecting wide variability across geographic and ethnic populations.[7]Among focal dystonias, the most commonly involved sites include the neck (cervical dystonia), eyelids (blepharospasm), oromandibular region, larynx, and limbs, particularly in task-specific presentations like writer's cramp or musician's dystonia.[6] In a study conducted in India, Prasad et al. reported upper limb dystonia as the most prevalent subtype (47.7%), followed by blepharospasm (26.1%) and oromandibular dystonia(18.2%).[8]

Beyond anatomical patterns, dystonia can also be classified by its temporal features as persistent, action-specific, paroxysmal, or diurnally fluctuating. Persistent dystonia remains all the time. Action-specific dystonias manifest only during tasks, paroxysmal dystonia appears episodically, while diurnal variation is typically seen in dopamine-responsive dystonia.[2,9] Diagnosis is primarily clinical and requires exclusion of mimics such as neuromuscular junction abnormalities, neck extensor myopathy, hemifacial spasm, and functional neurological disorders.[3,10]

While numerous international studies have explored the epidemiology and clinical characteristics of dystonia, such data from Pakistan remains scarce. A regional study reported that focal dystonias accounted for a significant portion of movement disorder referrals, yet sub type-specific data are largely lacking.[11] Incorporating local data into movement disorder research is essential not only for accurate diagnosis and management, but also for aligning with regional public health priorities.[12]

This study was therefore undertaken to describe the clinical spectrum of focal dystonia among patients attending neurology department of a tertiary care hospital in Karachi. By characterizing the prevalent sub-types and demographic patterns, the study aims to contribute to local data, enhance clinical recognition, and inform patient-centered management strategies in Pakistan's health-care setting. The objective of this study was to evaluate the clinical spectrum of patients with focal dystonia attending the neurology department at a tertiary care hospital in Karachi.

Methodology

This descriptive cross-sectional study was carried out at the Neurology Department of Dr. Ruth K. M. Pfau Civil Hospital, which is a tertiary care teaching hospital affiliated with Dow University of Health Sciences Karachi. The study was approved by the Institutional Review Board of the university on 18th of February, 2025 (Ref: IRB-3816/DUHS/Approval/2025/64). The study duration was six months, from 21st March 2025 to 20th September2025. Using WHO calculator, sample size of 75 cases was calculated with 95% confidence interval, 9% margin of error and percentage of oromandibular dystonia i.e. 18.18%.[13] This sample size was considered sufficient to describe the overall clinical spectrum of focal dystonia. Subsequent analyses of other focal dystonia subtypes (cervical, limb, blepharospasm, laryngeal) were performed descriptively, as the study was not powered to detect subtype-specific differences. We did not collect personally identifiable information such as names, addresses, email addresses, or phone numbers. To ensure anonymity, each participant was assigned a pseudonym. In order to maintain confidentiality all the paper based data was kept in areas where only investigators could access it . Electronic based data was saved in computers with limited access with password protection and regular backups. The inclusion criteria was participants between the age of 15 and 60 years of either sex, with focal dystonia. The exclusion criteria included patients with clinical conditions mimicking dystonia (neck extensor myopathy, post-traumatic deformities, Isaacs syndrome, neuromuscular junction disorders, and hemifacial spasm) and non-consenting patients. The participants were recruited using non-probability method of convenience sampling.

Eligible participants included both newly diagnosed with focal dystonia at the time of enrollment or had existing diagnosis prior to study entry. Diagnosis was determined by consultant neurologists with expertise in movement disorders, ensuring accuracy in recognition and classification. Focal dystonia was defined clinically as sustained, patterned, and often task-specific muscle contractions localized to a single body region. This aligns with established diagnostic frameworks and international movement disorder society guidelines.[2,3] After obtaining informed written consent, eligible patients were evaluated and data was recorded using a structured proforma capturing demographic information (age, gender, residence, education, occupation), clinical characteristics (age at onset, duration of illness, family history), and the specific type of focal dystonia. The spectrum of dystonia assessed included blepharospasm (involuntary spasm of orbicularis oculi muscles), oromandibular dystonia (involuntary movements of the jaw, tongue, or lower face), laryngeal dystonia (laryngeal muscle contractions resulting in dysphonia), cervical dystonia (involuntary twisting of the neck), and upper or lower limb dystonia. As temporal features (persistent, paroxysmal, and diurnal variation) reflect symptom behavior over time, classification was primarily based on patient-reported history, with clinical assessment at presentation used to corroborate task specificity, persistence, and fluctuation where demonstrable. This approach is consistent with established clinical practice and previously described classification systems for dystonia. Task-specific limb dystonias were explicitly screened during clinical assessment.

Data analysis was performed using IBM SPSS version 26.0. Continuous variables were reported as mean ± standard deviation, while categorical variables were reported in terms of frequencies and percentages. Statistical associations were explored using the Chi-square test, with a p-value ≤ 0.05 considered statistically significant. Given the exploratory nature of the study and small sizes of certain dystonia subgroups, results were interpreted cautiously.

Results

The study included 75 participants with a mean age of 32.09±11.68 years (95%CI:29.40–34.78), and the mean age at onset of dystonia was 29.02 ± 10.57 years (95% CI: 26.59–31.45). The average duration of illness was3.88±3.69 years (95%CI:3.03–4.73). The majority of participants were male (59, 78.7%), while 16 (21.3%) were female. A significant proportion of participants were from urban areas (89.3%), and 10.7% belonged to rural regions. In terms of educational status, 66.7% were illiterate, 22.7% had an education under matric level, and only 10.6% were graduates or above. Regarding employment, 57.3% were employed, and 42.7% were unemployed. Most participants had an active lifestyle (89.3%), while 10.7% reported a sedentary lifestyle. A family history of dystonia was present in 22.7% of the participants. Concerning the variability of symptoms, 77.3% experienced persistent dystonia, 10.7% had paroxysmal symptoms, and 12.0% reported diurnal fluctuation. No cases of focal dystonia in our study met criteria for task-specific presentation. Mean age differed significantly ($p<0.001$), being lowest in laryngeal (19.50±4.81years) and highest in cervical dystonia (39.25±10.76years). Duration of illness also varied ($p = 0.05$), with the longest in cervical (5.50 ± 4.58 years) and shortest in laryngeal dystonia (2.00 ± 1.06 years). Gender was significantly associated ($p<0.001$), with all blepharospasm patients being female and others predominantly male. Symptom variability showed significant differences ($p<0.001$): blepharospasm and oromandibular dystonia were entirely persistent, laryngeal was 50% persistent and 50% paroxysmal, while cervical presentation was 75% persistent and 25% had diurnal fluctuation.

Table 1: Association of Patient Characteristics with Clinical Spectrum of Focal Dystonia(n=75)

Patient Characteristics		Clinical Spectrum					P-Value
		Cervical Dystonia(n=28)	Limb Dystonia (n=18)	Blepharospasm (n=14)	Laryngeal dystonia (n=8)	Oromandibular dystonia (n=7)	
Age in years		39.25±10.76	29.00± 12.40	32.71± 6.91	19.50± 4.81	24.57±7.13	<0.001*
Duration of Illness in years		5.50±4.58	2.89±3.40	3.24±2.90	2.00±1.06	3.42±0.78	0.050*
Gender	**Male**	28(100.0)	16(88.9)	0 (0.0)	8(100.0)	7(100.0)	<0.001*
	Female	0 (0.0)	2(11.1)	14(100.0)	0 (0.0)	0 (0.0)	
Variability	**Persistent**	21(75.0)	12(66.7)	14(100.0)	4(50.0)	7(100.0)	
	Paroxysmal	0 (0.0)	4(22.2)	0 (0.0)	4(50.0)	0 (0.0)	<0.001*
	Diurnal fluctuation	7(25.0)	2(11.1)	0 (0.0)	0 (0.0)	0 (0.0)	

Discussion

This study provides a focused evaluation of the clinical spectrum of focal dystonia in a tertiary care hospital in Karachi, with particular emphasis on demographic patterns, subtype prevalence, and symptom variability. Cervical dystonia emerged as the most frequently observed sub-type in our cohort, accounting for 37.3% of cases, followed by limb dystonia (24.0%), blepharospasm (18.7%), laryngeal dystonia (10.7%), and oromandibular dystonia (9.3%) as shown in Table 1. This pattern differs somewhat from the findings of Prasad et al.[8] who reported upper limb dystonia as most prevalent in their Indian cohort, Similarly, Rajan et al. found that brachial dystonia accounted for 65.8% of cases, followed by cranial (27.1%) and cervical (15.7%) subtypes.[1] In contrast, results of our study are consistent with broader global trends suggesting increased recognition of cervical dystonia in clinical practice.[1,14]

The observed gender distribution in our study, where all blepharospasm cases were female and whereas other focal subtypes were predominantly observed in male patients (Table 1), contrasts with earlier reports that commonly describe a female predominance across most focal dystonia types, including cervical and cranial forms.[5,15] For example, a large European multicenter study reported a female-to-male ratio of nearly 2:1 in cranial and cervical dystonias.[16] This discrepancy may reflect regional sociocultural factors, differential care-seeking behavior, or referral biases rather than true epidemiological distributions. In our setting, male patients, who are often primary income earners, are more likely to seek neurological consultation when focal dystonia interferes with work performance, while cranial dystonias such as blepharospasm may prompt earlier consultation among female patients due to functional and cosmetic concerns. Notably, laryngeal dystonia showed the youngest mean age at onset (19.5years) , raising the possibility of early-onset idiopathic or hereditary forms, as supported by genetic studies in dystonia cohorts.[4,17] Young mean age at onset of laryngeal dystonia (19.5 ± 4.81 years), as shown in Table 1, is an intriguing finding from our study, which significantly differs from international data where onset is typically in mid-life (average 31–35 years).[18,19]

In terms of symptom variability, most cases were persistent, particularly among blepharospasm and cervical sub-types (Table 1), which is in line with findings from both SouthAsian and Western populations.[9,10] Moreover, recent reviews have underlined that focal dystonias, though relatively rare, remain under diagnosed and often misclassified, particularly in resource-constrained healthcare systems.[12] Interestingly, the average duration of illness prior to presentation varied significantly by subtype, with cervical dystonia patients having the longest mean duration (5.50 ± 4.58 years) as shown in Table 1. This finding reflects trends seen globally, where diagnostic delays are common, particularly for cervical and laryngeal dystonias.[18] One study reported an average diagnostic delay of over four years for laryngeal dystonia, often due to misdiagnosis as psychiatric or functional voice disorders.[18,19] This reinforces the need for broader awareness among non-neurologists, particularly ENT and primary care physicians, in identifying focal dystonias.

Another notable finding was the high proportion of illiterate (66.7%) patients, potentially reflecting disparities in access to care and information. This demographic skew warrants attention, as educational status may influence both care-seeking behavior and awareness of neurological symptoms, potentially contributing to delayed diagnosis and underreporting in rural settings.[13,19] A family history of dystonia was noted in 22.7% of our participants, comparable to reports from international registries and clinical cohorts.[20] This highlights the potential utility of developing national dystonia registries in Pakistan to support early identification of hereditary forms and contribute to global genomic research efforts.

The overall shortage of structured epidemiological data on dystonia in Pakistan reflects broader challenges in neurology infrastructure and research prioritization.[21] We advocate for enhanced clinician training, multidisciplinary collaboration, and public awareness campaigns to improve diagnostic accuracy and treatment outcomes. Botulinum toxin remains the mainstay of treatment for cervical and other focal dystonias; however, access and treatment outcomes may vary across regions due to resource availability and expertise.[22] Studies comparing treatment efficacy in European and Latin American populations emphasize the importance of localized treatment protocols and outcome monitoring.[22] In

the Pakistani context, such data are lacking, underscoring the need for research on therapeutic response and long-term management. The overall shortage of structured epidemiological data on dystonia in Pakistan reflects broader challenges in neurology infrastructure and research prioritization.[20] This study underscores the clinical utility of subtype classification in improving diagnostic accuracy and tailoring patient management. It supports the growing consensus that earlier recognition, broader awareness among clinicians, and local registry development are vital to advancing care for dystonia patients in developing regions.[6,16] Furthermore, understanding the distribution of focal dystonia subtypes will support timely referrals, reduce misdiagnosis, and guide effective treatment approaches.[16]

Limitations

Several limitations of this study warrant acknowledgement. First, the research was conducted at a single tertiary care facility, Dr. Ruth K. M. Pfau Civil Hospital, as an initial, exploratory effort to generate baseline data on focal dystonia in Pakistan, where subtype-specific data are currently scarce. This setting allowed for diagnostic consistency by expert neurologists within available time, resource constraints and ethical approvals. While this center serves a large and diverse patient population, the single-center design, modest sample size, and reliance on convenience sampling may introduce selection and referral bias. Consequently, the observed clinical spectrum may not be fully representative of the general population. The observed sex distribution across focal dystonia subtypes should be interpreted with caution. The predominance of females in blepharospasm and males in other subtypes likely reflects referral patterns, health seeking behaviour and center-specific sampling rather than true biological differences, hence the findings are not intended to represent population-level sex distribution. Additionally, the use of Chi-square testing in small subgroup analyses may have resulted in violations of test assumptions, and effect sizes or confidence intervals were not calculated. Therefore, statistically significant associations should be interpreted as exploratory and hypothesis-generating rather than definitive. Since the diagnoses were made by multiple clinicians without formal inter-rater reliability assessment, some degree of diagnostic variability cannot be excluded. One limitation of our study is that our assessment of cervical dystonia did not include documentation of its phenomenological subtype and directionality. Specific subtypes - including torticollis, retrocollis, laterocollis, anterocollis were not systematically recorded hence subtype specific comparisons could not be performed.

Despite these constraints, our findings contribute meaningful insights into the presentation of focal dystonia within the Pakistani context, where data on movement disorders remain scarce.

Conclusion

This study highlights cervical dystonia as the most prevalent form of focal dystonia in a tertiary care setting, followed by limb dystonia and blepharospasm. The findings underscore the clinical variability and demographic associations among sub-types. Recognizing these patterns can enhance diagnostic accuracy and inform targeted management strategies, particularly in regions with limited data on movement disorders like Pakistan.

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Data Availability Statement: The data that support the findings of this study, apart from the data already presented in the results section, are available from the corresponding author upon reasonable request

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