# IDIOPATHIC DYSTONIA

## CLINICAL, PROFILE OF 76 BRAZILIAN PATIENTS

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**SUMMARY — Dystonia may be classified by age of onset (childhood, adolescence, adult onset), body distribution of the abnormal movements (focal, segmental, unilateral, multifocal and generalized) and etiology (idiopathic and symptomatic). We studied 76 patients with idiopathic dystonia among 122; cases of dystonic syndrome (62.3% of the total). There were 48 female and 28 male patients. Adult-onset focal dystonia was the most frequent feature (37 patients). The onset of generalized dystonia was more frequently seen under the age of 20, whereas focal and segmental dystonia usually started over this age. Postural tremor of the hands was observed in 19.7% of the patients. Spasmodic torticollis was the most prevalent form of dystonia overall. Except for writer's cramp, which occurred more frequently in males, and generalized dystonia, which was equally divided between sexes, all other forms were more frequent in females. Our data suggest that differences in racial origin, social and economical status and environmental factors do not account for a different manifestation in dystonia pattern.**

**KEY WORDS: dystonia, dystonia musculorum deformans, idiopathic dystonia.**

**Distonia idiopática: perfil clínico de 76 pacientes brasileiros**

**RESUMO — A distonia pode ser classificada de acordo com a idade de início (infância, adolescência & idiade adulta), distribuição corporal dos movimentos anormais (focal, segmentar, unilateral, multifocal e generalizada) e etiologia (idiopática e sintomática). Dentre 122 pacientes com o diagnóstico de síndrome distônica, estudamos 76 com quadros idiopáticos (Gi\3% do total). Havia 48 pacientes do sexo feminino e 28 do sexo masculino. O quadro mais frequentemente observado foi a da distonia focal iniciada na idade adulta. (37 pacientes). Havia 6 pacientes com distonita generalizada e o início desse quadro foi mais frequento abaixo dos 20 anos de idade. Quadros focais e segmentares predominaram e foram mais comumentemente iniciados na idade adulta. Tremor postural das mãos foi observado em 15 pacientes (19>,7%). De todas as formas de distonia, o torcicolo espasmódico foi a que prevaleceu. Com a exceção da cãimbra do escrivão (com mais homens que mulheres acometidas) e da distonia generalizada (a mesma proporção entre os sexos), o sexo» feminino predominou sobre o masculino. Nossos dados são semelhantes aos de outras séries que estudaram o quadro clínico da distonia idiopática. Assim, diferenças raciais, ambientais e sócio-econômicas não parecem ser determinantes: no* padrão de manifestação da distonia idiopática.**

**PALAVRAS-CHAVE: distonia, dystonia musculorum deformans, distonia idiopática.**

**Oppenheim, in 1911, introduced the term dystonia to describe four patients presenting sustained abnormal postures and clonic or tonic muscle spasms in different parts of the body 32. Currently, the best accepted definition for dystonia is provided by the Ad Hoc Committee of the Dystonia Medical Research Foun-**

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dation (1984): "dystonia is a syndrome of sustained muscle contraction, frequently causing torsion and repetitive movements or abnormal postures" 9. According to this Committee, dystonia should be classified in terms of age of onset, body region affected and etiology. Anatomopathological studies failed to demonstrate basal ganglia lesions in idiopathic patients, but some showed neural loss and degenerative changes in brainstem nuclei[3]?. In symptomatic dystonia, lesions in the basal ganglia, especially in the putamen 6,14,2s; c a n ^ e found. This has led to the assumption that in idiopathic dystonia some sort of defective mechanism or biochemical abnormality is present. In a few patients neurotransmitter abnormalities have, indeed, been demonstrated

Under some specific circunstances treatment of dystonia may be successful 5,7,17,19. For this reason, a careful scrutiny of the cases, and knowledge of the clinical characteristics of the various forms of dystonia is necessary. To improve our ability to make this assessment in our environment, we decided to study the clinical profile of 76 patients with this diagnosis.

## PATIENT'S AND METHODS

We studied the notes of patients* with the diagnosis of dystonia syndrome as defined by the Ad Hoc Committee of the Dystonia Medical Research Foundation 9. All these patients had been evaluated in the Sector for Investigation of Extrapyramidal Diseases of Eseola Paulista de Medicina or in one of our private practices, between January-1932 and December-1988.

We decided to exclude the patients under 12 years of age at the time of their first assessment and those with parkinsonism. Patients whose disease began before 12 years of age but were seen when older were included in the study. Patients whose notes were incomplete were also excluded. The diagnostic criteria for idiopathic dystonia were a normal neurological examination (except for the dystonia) and no definable cause of the syndrome aside from genetic predisposition. Blood tests and computerized tomography (CT) of the brain were performed when necessary, and their results had to be normal. The single hemidystonia patients had a normal CT scan, but no MRI study was available.

Of 122 dystonie patients, 76 fulfilled the diagnostic criteria for idiopathic dystonia, whereas the other 46 symptomatic dystonie patients are being presented in a separate paper *12*. The sex, racial origin, age of onset and the body distribution of the dystonia, were studied. The occurrence of tremor along with the dystonia was also noted.

## RESULTS

Idiopathic dystonia accounted for 62.3% of the total group. The sex distribution and the mean lage of onset are shown in Table 1. There were two patients with a positive family history of dystonia (2.6%). They were both female. The first had Meige syndrome which started at age 67, and had two brothers with dystonie syndrome, one of them with spasmodic torticollis and the other suffering from idiopathic blepharospasm. The second patient had generalized dystonia which began at age 15. Her father and two of her four siblings had a similar generalized clinical picture. In none of the affected relatives was a neurological verification possible.

Table 1. Distribution of patients according to age of onset (mean) and sex.

| | Nº | % | Age of onset (in years) |
|---|---|---|---|
| Male | 28 | 36.8 | 36.2 |
| Female | 48 | 63.2 | 38.2 |
| Total | 76 | 100 | 37.5 |

As far as the racial origin was concerned there were just three patients of Jewish ancestry. They were all of the female sex and none had ta positive family history for dystonia. Two suffered from writer's cramp and the third from oromandibular dystonia.

It is shown in Table 2 the age of onset in relation to the body area distribution of dystonia. The data shows that focal and segmental dystonia tended to start at an adult age whereas the generalized dystonia started in childhoood and adolescence.

**Table 2.** Distribution of patients according to age of onset and affected body area.

| | Childhood (0-12 years) | | Adolescence (12-20 years) | | Adult (> 20 years) | | Total |
|---|---|---|---|---|---|---|---|
| | No. | % | No. | % | No. | % | No. |
| Focal | 1 | 2.2 | 7 | 15.6 | 37 | 82.2 | 45 |
| Segmental | 1 | 4.5 | 2 | 9.1 | 19 | 86.4 | 22 |
| Unilateral | — | — | — | — | 1 | 100.0 | 1 |
| Multifocal | — | — | 1 | 50.0 | 1 | 50.0 | 2 |
| Generalized | 1 | 16.7 | 4 | 66.7 | 1 | 16.7 | 6 |
| Total | 3 | 3.9 | 14 | 18.4 | 59 | 77.6 | 76 |

The freciuency of each classical form of dystonia as well as the frequency for each sex and the mean age of onset are depicted in Table 3. Spasmodic torticollis, essential blepharospasm (including cased of Meige syndrome) and writer's cramp were the more prevalent forms. The female sex predominated in those having spasmosdic torticollis, Meige syndrome (including cases of essential blepharospasm and oromandibular dystonia.) and spastic dysphonia. The opposite occurred in the patients with writer's cramp. Patients with Meige syndrome had the latest onset (56.5 years), whereas those with generalized dystonia had the youngest onset (17.2 years). Spasmodic torticollis and writer's cramp mostly started around 30 years of age.

In 15 patients postural tremor in the hands was noted (19.7%). Myoclonus was noted in two other patients.

**Table 3.** Clinical profile of patients considering the frequency of sex and mean age of onset.

| | No. | % | Sex Male/Female | Mean age of onset (in years) |
|---|---|---|---|---|
| ST | 25 | 32.9 | 4/21 | 29.0 |
| isolated | 13 | | 1/12 | 30.2 |
| associated | 12 | | 3/9 | 25.9 |
| Writer's cramp | 20 | 26.3 | 12/8 | 34.5 |
| Meige syndrome | 12 | 15.8 | 4/8 | 56.5 |
| OMD | 18 | 23.7 | 4/14 | 50.9 |
| isolated | 1 | | 0/1 | 54.0 |
| other non-Meige associations | 5 | | 0/5 | 37.0 |
| Blepharospasm | 22 | 28.9 | 8/14 | 52.0 |
| isolated | 8 | | 4/4 | 50.9 |
| other non-Meige associations | 2 | | 0/2 | 29.5 |
| SD | 7 | 9.2 | 2/5 | 41.6 |
| isolated | 3 | | 1/2 | 48.7 |
| associated | 4 | | 1/3 | 36.2 |
| Generalized | 6 | 7.8 | 3/3 | 17.2 |

*ST, spasmodic torticollis; OMD, oromandibular dystonia; SD, spastic dysphonia.*

## COMMENTS

**In this retrospective study a detailed description of the patients is not possible. A follow-up study of the patients concerning the temporal modifications in the presentation of dystonia was also impossible to obtain. A large number of patients attended on an insufficient number of visits to allow a longitudinal study. Patient compliance in this country is affected by cultural and economic factors, including difficulty in access to proper medical care and this might have played a role in our series. Patients aged below 12 in our first evaluation were excluded as they are seen in the Child Neurology Division. As these patients are examined by other neurologists, a difference in evaluation could lead to erroneous results. Also, we are trying here to present the experience of a Sector of Investigation in Extrapyramidal Disease working with non-pediatric patients.**

**A greater frequency of female patients in idiopathic dystonia had already been observed by others but, perhaps not to the same extent as here. A 1/1.1 male/female ratio was found by Marsden and Harrison 27, but cases of spasmodic torticollis (ST) were deliberately excluded by them. Most of our cases of ST were women (Table 3) and this may have enlarged the overall female proportion (63.2%).**

**Familial cases were seen in a small number of patients (2.6%) and this figure is substantially different from what is commonly found. Marsden et al.[28] found 22% of familial cases in their series. When just generalized forms of idiopathic dystonia are considered, an even greater proportion of hereditary cases are seen (24%) 8. Many reasons may be put forward to explain our low frequency of familial cases. The exclusion of patients below 12 years of age may be in part responsible. It is well known that the earlier the onset of the dystonia, the greater the chance of it becoming generalized and of being hereditary 8,28. Most of our cases consist of people in the lower social classes, many of whom are migrants from distant parts of the country who are away from their families for a long time. Also, "formes frustes" of the syndrome appearing in their distant relatives may pass unnoticed to them 36 in addition, these "formes frustes" may not be given sufficient consideration as a disease manifestation. The low frequency of Jewish ancestry of our patients could have also contributed to the small number of familial cases. Although the clinical picture of idiopathic dystonia is not different between Jews and non-Jews[4], heredity plays a more considerable role in the formers.**

**Dystonia beginning in childhood and adolescence had the tendency to be generalized, as seen in Table 2. Fahn et al. n observed that more than 80% of the cases of dystonia in these age groups are of generalized distribution. Childhood and adolescent-onset dystonia usually begin focally and have a 75% chance of progressing slowly to become generalized ^. In a study of 116 childhood-onset dystonic patients it was noted that when dystonia started in a leg there was a 89% chance of it becoming generalized; when it started in an upper limb, this possibility would be 70%; and in the neck muscles, 33%-is. Over the age of 20, focal and segmental dystonias largely predominated. In the study performed at Columbia-Presbyterian Center of New York by Fahn et al. n, where 625 idiopathic dystonia patients were seen, symptoms began after 20 years of age in 69%, whereas in our study this was the case in 77.6%. Our higher frequency of adult-onset cases is likely to be due to the exclusion of the pediatric population.**

**The simultaneous occurrence of tremor and dystonia was already been pointed out by many authors 1,21,33,27,35. This tremor manifests itself in the same form as the essential tremor 21. Whether this is a coincidental occurrence of two separate entities or not is still in debate. The frequency of postural tremor may reach up to 14% in various forms of idiopathic dystonia 27. in isolated forms of spasmodic torticollis (ST) tremor is observed in 33% of the patients. In our total group of idiopathic dystonia the frequency was 19.7% which is similar to what we found in our group of symptomatic dystonia[12].**

**The most frequent clinical presentation of idiopathic dystonia was ST (32.9%), considering focal or segmental and multifocal forms. If we compare just the focal presentation of dystonia, writer's cramp (WC) is the most frequent (26.3%).**

**Spasmodic torticollis — ST may be defined as the result of involuntary contractions of the neck muscles, leading to sustained deviation of the head, which may or may not be associated with dystonia in another body segment[1] Many of our patients performed a peculiar trick which was useful in reducing or abolishing the muscular contraction by touching the chin, the face or the back of the head. In an epidemiological survey performed in Rochester (USA), ST was the most prevalent focal dystonia (89 cases per 100 000 inhabitants) and showed the greatest incidence (11 cases per 100 000 inhabitants per year) si. The female predominance in ST has been reported by many authors. Friedman and Fahnis report a male/female ratio of 1/1.6 in the isolated forms of ST and a 1/1.3 ratio when the associated forms were considered. Others have sex ratio of 1/1.4 up to 1/4.515,23,31, CHi patients show an overall ratio of 1/5 in favor of women with the greatest difference in the isolated form (1/12) compared to associated forms (1/3). We cannot offer a satisfactory explanation for this larger female predominance in our patients. Perhaps the smaller number of patients of our series may account for it. The mean age of onset for the total group of ST (29 years) is slightly lower but still comparable to other papers, whose mean ages ranged from 32 to 45 years 13,15,23,31.**

**Writer's cramp (WC) — All 20 WC patients of this study were isolated forms of focal dystonia. During handwriting, a spasmodic contraction in the muscles of the hand and forearm occurs, producing deviation of the wrist, difficulty in controlling hand movements and frequently, pain in the affected muscles. The pen may be held in an awkard position and excessive force is used to keep the pen on the surface of the paper. As the subject begins to write, the wrist and elbow raise, the forearm is pronated and the fingers extend. This task specific dystonia may be produced by activities other than handwriting, such as typewriting and working with a computer keyboard28, playing musical instruments[22], or sporting activities, such as playing golf so. The generic term, occupational dystonia s[4] has been put forward to encompass these types of task specific dystonia. All our patients had the classical form of handwriting dystonia (WC). In the USA the incidence has been estimated to be 3 cases per 100000 inhabitants per year 31. The male preponderance (60%) observed here is in agreement with other studies. Marsden reported that of 36 patients with WC, 66% were male and their mean age of onset was 47 years in isolated forms and 33.8 years in the associated forms[24]. This figure is similar to our patients (34.5 years). Nutt et al.si observed a mean age of onset of 49 years.**

**Meige syndrome (MS), essential blepharospasm (KB) and oromandibular dystonia (OMD) — EB and OMD were both frequent in our study, either in isolated or associated forms. The association of both dystonic syndromes occurring spontaneously in a single patient is known as Meige syndrome (MS). Isolated EB occurred in 8 cases while associations other than MS accounted for 2 cases, A total of 22 cases was observed including MS and the other forms of blepharospasm. Isolated OMD was seen in a single patient and associated forms other than MS in 5 cases. The total number of cases of OMD was 18, while pure MS was present in 12 patients. Marsden 25 collected 39 patients with these presentations of dystonia, 13 with isolated blepharospasm, 9 with isolated OMD and 17 with MS. EB is characterized by simultaneous spasmodic contractions of the orbicularis oculi muscles. Tricks, such as opening the mouth widely or producing sounds may enable the patients to open their eyes. This decrease in intensity of the dystonia produced by voluntary movements of cranial muscles is called the paradoxical phenomenon w*. One of our patients developed this trick. According to Nutt et al. the annual incidence of EB is estimated as 5 cases per 100000 inhabitants si. Overall, blepharospasm was more common in women. The same occurred with the OMD and MS cases, and this supports the findings of other studies 2fc3i. i n a m o r e recent review, Marsden found his cases of MS equally divided by sex26. The mean age of onset of all these forms of cranial dystonia was around 50 years which is similar to other reports 25,26,31.**

**Spastic dysphonia (SD) — Spastic dysphonia was responsible for 9.2% of our series of idiopathic dystonia. Blitzer et al.[2] found 110 cases of SD among 1280 dystonic patients (£.6%). SD defines the voice and speech disturbance produced by laryngeal dystonia where the laryngeal adductor musculature in impaired in its more frequent presentation (adductor dysphonia). In a smaller**

proportion of patients the abductor muscles are impaired (abductor dysphonia). In the former type the voice is spastic and interrupted; in the latter the patient seems to whisper, and is frequently thought to have psychological dysfunction 33. All our cases of SD were of the adductor type.The female predominance and the age of onset around 40 years is similar from other studies[2,3].

This is the first clinical characterization of idiopathic dystonia in Brazilian patients. Previous series have referred mainly to North-American or European populations that very different to our patients in terms of social, economic and cultural status. The rich mixture of races and the different environment of our country are additional factors which could have some importance m the disease pattern. Notwithstanding, once dystonia manifests itself, the clinical pattern will be the same as of other populations. Further epidemiological surveys are necessary to establish the incidence and prevalence rates in our country to allow a comparison with other parts of the world.

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