Frequency of Primary Torsion Dystonia for the Population of Bulgaria – A Service-Based Survey

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Abstract: *Background*: According to the majority of epidemiological studies for primary torsion dystonia (PTD), it is a rare disease, although there are some controversial data, showing much higher figures. Because the need of clarification in this field, the aim of this study is to estimate the frequency of primary torsion dystonia in the Bulgarian population. *Materials and Methods*: We made an official inquiry to the National Center for Health Information, and to the National Health-Insurance Fund for the total count of all the patients, registered with the diagnose of PTD for a period of a year. Simultaneously we examined the archive of the University Multidisciplinary Hospital for Active Treatment of Neurology and Psychiatry - UMHATNP "St. Naum" in Sofia, with a specialized department of Movement Disorders, for the period of 01.01.2008-01.01.2012. *Results*: Crude annual prevalence of primary torsion dystonia estimated in this study is significantly lower than most reported epidemiological findings on prevalence. Probably the actual prevalence is higher than that calculated in the current study, but this could be demonstrated by improving the collection of data and methods for screening and diagnosis. *Conclusions*: The downside of this study, as with most studies in this area worldwide, is that it is based on the demand of medical care. Thus remain not estimated all undiagnosed and and incorrectly diagnosed cases and patients that have not sought medical attention. The majority of the patients in our sample came from the South-Western Region of the country. According to distribution by form of dystonia, age, gender and time to diagnose our results with some exceptions are similar to those, reported in the literature.

Keywords: Bulgarians, dystonia, prevalence.

INTRODUCTION

Dystonia is a movement disorder, characterized by intermittent or sustained simultaneous muscle contractions of opposing muscles resulting in twisting and repetitive movements, abnormal posture or both. When no etiologic factor can be identified, the dystonia is referred to as primary torsion dystonia (PTD) [1]. PTD is usually provoked or exaggerated by voluntary movements and it could be accompanied by tremor [2, 3]. The disorder may affect any part of the body – neck, face, trunk, extremities [1, 4] often causing serious disability. The disease shows great variability according to age of onset (early or late onset), etiology (variable gene expression and penetration, sporadic forms), affected body areas (focal – cervical, blepharospasm etc.; segmental; multifocal; generalized) and severity (varying from mild forms, manifesting only with specific voluntary movement to severe dystonic "storms" causing life-threatening myoglobinuria) [1, 5, 6]. This variability of the condition may introduce some difficulties in estimating PTD frequency [4]. Not to underestimate the fact that mild and early forms are often hardly recognized or might be misdiagnosed [4, 5]. According to the majority of epidemiological studies for primary torsion dystonia, it is a rare disease [5, 7, 8], although there is wide spread of the frequency data [5, 9], and a final assessment needs more researches in this field. The aim of this study is to estimate the frequency of primary torsion dystonia in the Bulgarian population.

MATERIALS AND METHODS

To investigate the frequency of PTD in Bulgaria we made an official inquiry to the National Center for Health Information, where we found no official data published for the prevalence and incidence of primary torsion dystonia in Bulgaria.

Then we made an official request to the National Health-Insurance Fund for the total count of all the patients, registered with the diagnosis of PTD for a period of one year – respectively 2010 and 2011. There were 52 patients with PTD registered in 2010 and 55 in 2011.

Simultaneously we examined the archive of the University Multidisciplinary Hospital for Active Treatment in Neurology and Psychiatry- UMHATNP "St. Naum" in Sofia, with a specialized department of Movement Disorders, for the period of 01.01.2008-01.01.2012.For the four years period there were hospitalized altogether 103 patients with a preliminary diagnosis of PTD– 30 patients in 2008; 31 patients in 2009; 18 patients in 2010 and 24 patients in 2011. In 19 patients were found symptoms or instrumental findings indicating secondary dystonia, or the diagnosis was uncertain and they were excluded from further analysis. There were 12 patients with more than one

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hospitalization during this period and subsequent hospitalizations were excluded from the analysis. A total of 84 patients had a confirmed diagnosis of PTD.

In 2011 a national census was carried out with an estimate for the population of 7364570 persons which we used in calculating crude annual prevalence.

All the data was statistically analyzed by using the SPSS program, assuming a level of significance $\alpha = 0.05$. Descriptive statistics was performed and the crude annual prevalence was estimated within 95% CI.

RESULTS

The crude annual prevalence of PTD in the country in 2011 was estimated as the whole number of old and new cases for the year (55 cases) divided to the old cases, new cases and non-affected people which were equal to the entire population of the country for the estimated period -7364570 persons. So the crude annual prevalence of PTD for 2011 was estimated to 0.00074681889%, with 95% CI [-5.4944e-6 + 9.4419 e-6] or less than 0.001% or 7.5/1 000 000 in our service-based survey.

During the years 2010 and 2011 there were respectively 18 and 24 patients with PTD hospitalized in UMHATNP "St. Naum", and all the registered cases in the country were 52 in 2010 and 55 in 2011. This means that about 1/3 of all registered cases per year were hospitalized in UMHATNP "St. Naum" and this gives us a reason to assume that the following analyses of the patients with a confirmed diagnosis of PTD, hospitalized during the mentioned four years period, might be representative for this part of the country population who searched medical care.

From all 84 patients with confirmed diagnosis PTD females were 66.7%, and males – 33.3%. All the examined patients were over the age of 18, between 23 - 82 years old with mean age of 53.7 years (SD=16.4)

The majority of these patients were with focal dystonia – 72.6%, and the most of them were with cervical dystonia – 41.7%. From the other forms of focal dystonia there were patients with blepharospasm – 10.7%, oro-facial dystonia – 10.7%, and with focal hand dystonia – 9.5%. Patients with segmental dystonia were 17.9%. Patients with multifocal dystonia were 2.4%, and those with generalized dystonia – 7.1%.

Female gender was prevalent in all forms of focal dystonia (72%). In cervical dystonia females were 71.4%, in blepharospasm and in oro-facial dystonia – 77.8%, in focal hand dystonia – 62.5%. In patients with segmental dystonia there were slight predominance of female gender again – 60%. In patients with multifocal dystonia the two genders were equally presented, based on two cases, but in generalized dystonia there were predominance of male gender – 66.7%.

Time from the beginning of the first complains to diagnose the disease was about 4.1 years (SD=6.2). In 49.3% PTD was diagnosed until the end of the second year of the beginning of the complains, and 13 % of the cases were diagnosed after 10 or more years. The time required to diagnose different forms of PTD is presented in Table **1**.

According to the mean age of onset the focal forms of dystonia began about the age of 48.5 years (SD=15.6). The age of onset of cervical dystonia was about 43.7 years (SD=14.9), of blepharospasm – about 59.7 years (SD=7.6), of oro-facial dystonia – about 59.8 years (SD=15.3), and of focal hand dystonia – about 48.7 years (SD=17.1).

The majority of the patients – about 40%, were from the capital - Sofia. Most patients came from the South-Western Region of the country (the capital is located in the same region) – 58.6%, followed by the patients from the South-Eastern Region – 13.3%. The patients from the South-Central Region were 11.9%. The patients from the North-Western Region were 8%, from the North-Eastern Region – 4%, and from the North-Central Region – 3.9%.

DISCUSSION

For primary dystonia, precision of both incidence and prevalence estimates is problematic because of

Table 1: Time, Required for Diagnosis of Different Forms of PTD

Dystonia	generalized	multifocal	segmental	focal	cervical	blepharospasm	oro-facial	focal hand
Time to diagnose	2.5 years (SD=3.0)	5.7 years (SD=5.7)	4.9 years (SD=5.8)	4.2 years (SD=6.6)	4.7 years (SD=7.3)	5.4 years (SD=7.2)	0.8 years (SD=1.2)	2.8 years (SD=4.5)

the small number of cases identified in published, service-based studies [5]. The disadvantage of this study, as most studies in this area worldwide, is that it is based on the demand of medical care. Thus remain not estimated all undiagnosed and incorrectly diagnosed cases, and patients that have not sought medical attention (for example some of mild and early forms of the disease), and also most uninsured persons. With this type of epidemiological studies increases the risk of error in assessing the morbidity indicators, and comparing different subgroups of patients [4, 5, 10].

For the period 1996-1997, in eight European countries brute annual prevalence was estimated to be 152/1000 000 [11]. Prevalence of primary torsion dystonia in Bulgaria was approximately 7.5/1 000 000, making it significantly lower than most reported epidemiological findings on prevalence. Probably the actual prevalence is higher than calculated in the current study, but this could be demonstrated by improving the collection of data by using community-based methods andscreening questionnaire followed by expert examination for confirmation of the diagnosis.

The majority of authors consider focal dystonia to be the most common form of primary torsion dystonia [10, 11], and cervical dystoniahas a major share ofall focal forms [4]. The results obtained from this study do not differ from the literature.

According to the epidemiological evidences for Europe female gender is predominant in all forms of primary dystonia, except graphospasm [11, 12]. Our results revealed predominance of females in focal and segmental forms, men and women were equally represented (based on two cases) in multifocal dystonia, and a prevalence of male gender was found in generalized dystonia.

The age of onset of the different forms of focal dystonia in our study was very similar to those reported in most European countries, Japan, India, Egypt [5, 13-16].

In the United States the average period from the onset to establishing the diagnosis of primary torsion dystonia takes in average 4-8 years [17]. About 4.1years (SD = 5.2) were required for diagnosing the condition in the examined sample.

In this study most of the patients were from Sofia. This could be explained on one hand, with the demo-graphic distribution of the population, concentrated mainly in and around the capital, and on the other hand - with the location of UMHATNP "St. Naum" in Sofia, which may lead to increased rates of attendance by inhabitants of the city.

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