Spasmodic torticollis – a multicentre study on behavioural aspects II: signs, symptoms and course

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This paper deals with signs, symptoms and course in spasmodic torticollis (ST). Two hundred and fifty-six patients were included in the study, 59.3% women, 40.7% men. The mean age was 49.1 years. Rotating torticollis out-numbered latero- and antero-retrocollis. A family history of ST occurred in 3.1% of the total sample. First degree relatives were affected in 2.3%. Thirty-four per cent of the patients had additional dystonic symptoms. Most frequently these affected the upper extremities (13%), and less often the legs. Of the patients 19.1% had experienced a period of complete remission. The correlations between the severity of the signs and the neurological symptoms are surprisingly weak.

Keywords: Spasmodic torticollis – Family history – Signs – Symptoms – Course

INTRODUCTION

This paper deals with the prevalence of the neurological signs and their interrelations with symptoms, which was a question outlined in a previous paper (Scheidt *et al.*, 1996).

In the absence of definite knowledge on the aetiology and the underlying pathophysiological mechanisms, the diversity of the clinical picture in spasmodic torticollis (ST) has given rise to the question whether subgroups can be differentiated (Meares, 1971; Schulze and Stephan, 1987; Duane, 1988a). In addition to the predominant deviation of the head, characteristics such as the course of the illness, additional dystonic symptoms and a family history of dystonic movement disorders may serve as clinical criteria. These will be addressed in this paper.

Furthermore it will be reported in detail which clinical features are likely to be associated with psychosocial changes and psychopathology. Pain for example, if present, may particularly influence psychological well-being. Similarly deviation of the head is often more debilitating if it is phasic rather than tonic (Duane, 1988a). As will be illustrated, the

degree of distress due to the neurological condition varies considerably between patients. The physical signs, however, are not powerful predictors of subjective distress.

RESULTS

Sex-ratio, age and age of onset

Of the 256 patients, 152 were female (59.3%) and 104 male (40.7%) with an average age of 49.1 years (SD = 13.1) and an age range between 19 and 81 years. The mean duration of the disease was 8.9 years (SD = 7.9) ranging from 8 months to 54 years.

The mean age at onset differed significantly between the sexes ($p < 0.01$) (Fig. 1). It amounted to 40.9 years for the total sample (SD = 13.3 years), but was 42.9 years for women and 38.1 years for men. No significant differences between the sexes emerged with regard to the duration of symptoms and severity ([TSUI]-index).

FIG. 1. Age at onset related to sex.

Signs

Rotation, laterocollis and antero/retrocollis describe the predominant deviation of the head, but were not mutually exclusive. Therefore to register the complex three-dimensionality of the deviant head position, an individual evaluation of the head position in all three axes was made (Scheidt *et al.*, 1996). The results are shown in Fig. 2.

Rotation (deviation of the head in the horizontal plane) was the most frequent symptom occurring in 95% of the patients. Laterocollis (deviation of the head in the vertical plane) ranked second in frequency with 74.6%. Antero-retrocollis (inclination in the sagittal plane) occurred in 64%.

In 67.2% the abnormal head position was combined with tremor. Neither the tremor of the head nor the postural abnormality according to the patient's assessment improved in response to alcohol.

The mean score of the TSUI-index for the total sample amounted to 10.2 (SD = 3.40). Thirty-four per cent of the patients had dystonic symptoms other than torticollis. Most frequently these affected the upper extremities (13%), less often the legs (in 6% the right leg, in 8% the left leg). The trunk was

involved in 6%. The existence of additional dystonic symptoms did not correlate with severity. The TSUI-index in the multiple dystonic symptom group amounted to 10.9, compared to 9.8 in patients with isolated cranial dystonia. A number of other measures, however, such as the duration of the illness, the psychosocial changes and psychopathology differed between the two groups. Because they refer to psychosocial aspects they will be discussed in the fourth paper in this series (Scheidt *et al.*, 1996).

Symptoms

The degree of subjective distress due to the neurological symptoms, rated on a scale from 0 to 4 (Scheidt *et al.*, 1996), yielded the following mean values: tension of the head muscles 2.86, SD = 0.99, deviation of the head 2.56, SD = 0.98 and pain 2.15, SD = 1.21 (Fig. 3).

Sixty-six per cent of the total group felt severely or very severely disturbed (categories 3 and 4 of a scale ranging from 0 to 4) by the tension of the neck muscles, 62.7% by the deviation of the head. Pain and tremor of the head caused less frequent distress. Of patients suffering from pain 86% indicated that

FIG. 2. Deviation of the head in relation to the neurologist's assessment. Each patient was rated in three dimensions.

the symptom was progressive. Muscle tension, tremor and pain did not correlate with age, sex, severity or the duration of the illness.

History

For the majority of patients (53.0%), ST began with "twinges" and "tension" of the neck muscles. Less often the onset was characterized by a noticeably deviant position of the head (38.3%), pain (30.4%), or head tremor (24.5%). Three and a half per cent of the patients had had proven cervical spine trauma within 14 days of the onset of the illness. An additional 9.3% had other accidents in the same period of time, in which a trauma of the cervical spine area may have occurred, but could not definitely be proven.

Of the total sample, 107 patients (41.6%) had first seen a general practitioner for their symptoms. Eighty-eight patients (32.9%) had consulted a neurologist immediately. Other specialists, to whom the patients had been referred were the following: orthopaedic surgeons (15.9%), internists (5%) and others (2%).

The time from the first visit to a physician to the definite diagnosis of ST averaged 1.8 years. In the course of the disease, a mean of 6.4 physicians (SD = 5.5) were consulted for treatment.

Family History

A family history of ST occurred in 3.1% of the total sample. First degree relatives were affected in 2.3%, second degree relatives in 0.8%.

A tremor of the head or of the upper extremities in first or second degree relatives was reported in 9%. A further 5.1% of the patients reported a family history of Parkinson's disease. In total, 17.2% of the patients reported a positive family history of extrapyramidal motor symptoms. The positive family history of extrapyramidal motor disease did not correlate with age, sex, duration or severity of the torticollis.

Course

Of the patients 19.1% experienced a period of complete remission with an average length of approximately 1.1 years. Remission was more likely to occur in patients with a younger age of onset. The mean age of onset in the remission group was 37.2 years compared to 41.8 years in the non-remission group ($p < 0.05$).

Interrelations of signs and symptoms

Signs and symptoms were highly interrelated. The TSUI-index correlated significantly with rotation, laterocollis and antero-retrocollis (Table I). A negative

FIG. 3. Self-reported neurological symptoms, mean scores and standard deviations.

TABLE I. Correlations between socio-demographic variables, signs (neurological assessment) and symptoms (self-reported). The correlations between signs and symptoms are weak

	Socio-demographic variables			Neurological signs				Neurological symptoms			
	1	2	3	4	5	6	7	8	9	10	11
1 Age	—	0.80***	0.26***	0.05	-0.05	0.05	0.12*	0.11*	0.05	0.06	0.04
2 Age at onset	—	0.25***	0.11	0.04	0.04	0.13*	0.17**	0.11*	-0.00	0.09	
3 Duration			—	-0.11	0.14**	-0.00	-0.02	0.01	-0.02	0.15**	-0.04
4 TSUI-index				—	0.21**	0.47**	0.35***	0.13*	0.08	0.06	0.10
5 Rotation					—	0.16**	-0.24***	-0.14*	-0.08	0.06	-0.05
6 Vertical inclination						—	0.02	0.16*	0.04*	0.12	0.08
7 Sagittal inclination							—	0.14*	0.13*	0.04	0.11
8 Deviation head								—	0.54***	0.27***	0.42***
9 Muscle tension									—	0.19**	0.66***
10 Tremor of head										—	0.21***
11 Pain											—

* $p < 0.05$; ** $p < 0.01$; *** $p < 0.001$.

correlation emerged for the measures of rotation and antero-retrocollis. However, this might be a measurement artefact because antero-retrocollis in pressure rotation is difficult to assess. The finding might also correspond to the clinical observation that antero-retrocollis and rotation are only rarely associated, whereas rotation and vertical inclination often are.

The symptoms were also interrelated. For example, deviation of the head correlated with muscle tension

($p < 0.001$), tremor ($p < 0.001$) and pain ($p < 0.001$). However, the correlations between signs and symptoms were weaker than expected. Distress due to the deviation of the head correlated only weakly with the TSUI-index ($r = 0.13$, $p < 0.05$), rotation ($r = -0.14$, $p < 0.05$) and antero-retrocollis ($r = 0.14$, $p < 0.05$). (The negative correlation between rotation and distress due to the deviation of the head may result from the negative correlation between

TABLE II. Forms of treatment and their effects according to the patients' assessments. Treatment with botulinum toxin A is indicated as the most effective form of treatment

Treatment	No. of patients	Patients receiving therapy (%)	Effects of treatment (% of patients)		
			Better	Unchanged	Worse
Drug therapy					
Botulinum toxin A	179	69.8	83.2	12.0	4.8
Systemic drugs	205	80.2	27.9	58.0	14.1
Homeopathic medication	62	24.1	11.8	80.4	7.8
Surgery					
Peripheral neural dissection and rhizotomy	21	8.1	33.3	46.7	20.0
Muscle dissection	11	4.3	33.3	44.4	22.3
Stereotactic surgery	19	7.6	46.6	26.7	26.6
Decompression of the accessory nerve	7	2.9	33.3	33.3	33.3
Others					
Physical therapy	184	71.8	34.4	54.5	11.1
Chiropractic treatment	62	24.1	10.0	70.0	20.0
Psychotherapy	93	36.3	29.9	58.4	11.7
Hypnosis	26	10.0	19.0	71.4	9.6
Acupuncture	80	31.1	7.9	84.1	8.0

rotation and antero-retrocollis. The latter largely accounts for the distress due to the deviation of the head.) The correlation between distress due to tension of the neck muscles and vertical or sagittal inclination was also weak (Table I).

Treatment

Therapeutic diversity. Thirty-seven per cent of the patients had tried 1–3 different forms of treatment for ST, 40% between 4–6 and 15% more than 7. Only 8% of the patients had received treatment with botulinum toxin only.

The most common forms of treatment were systemic drugs (neuroleptics, anticholinergics and reserpine-analogues), physical therapy and botulinum toxin followed by psychotherapy and acupuncture (Table II). A total of 19 patients (7%) underwent surgical procedures.

Treatment with botulinum toxin. According to the patients' assessment local injection of botulinum toxin was the most effective treatment leading to symptomatic benefit in 83% of the cases (Table II). Improvement of the neurological condition was reported to have beneficial effects in different areas of life. Of the patients 76.8% felt that the treatment had improved their professional situation and 71.8% indicated an improvement in their social relationships (Fig. 4).

The mean duration of the effects of the treatment with botulinum toxin varied considerably. In 13% it lasted only up to 1 month, in 24.7% up to 2 months.

In many patients (48.8%) the injection held up to 3 months. The mean duration of the effects was 2.6 months.

DISCUSSION

Compared to other studies on large ST samples (Duane, 1988a,b; Jahanshahi and Marsden, 1988; Chan *et al.*, 1991; Rondot *et al.*, 1991), the patients described are similar with regard to the age at onset (40.9 years), the sex ratio (female to male 1.5:1) and the pattern of the predominant head deviation. Rotation is the most prevalent sign (Chan *et al.*, 1991; Rondot, 1991; Deuschl *et al.*, 1992). We found an earlier age of onset of ST in men than has been reported in the literature (e.g. Duane, 1988a,b).

The onset of ST is often non-specific with tension of the head muscles and a gradually developing deviant head position. The time from first presentation to a physician to the determination of the definite diagnosis amounted on average to 1.8 years. Considering that ST is a defined neurological disease this suggests a widespread diagnostic uncertainty. In our view neurological specialists should contribute to improve the diagnosis and treatment of ST in the primary care system.

Thirty-four per cent of the patients suffered from dystonic symptoms other than torticollis. These most frequently affected the upper extremities. The relatively high prevalence of additional dystonic symptoms in our study corresponds to numerous reports in the literature (Herz and Hoefer, 1949; Couch,

FIG. 4. Patients' assessments of improvement in their professional and social lives as a result of treatment with botulinum toxin A.

1976; Chan *et al.*, 1991). Rondot *et al.* (1991) reported that almost half of their sample complained of various degrees of difficulties in writing or impaired dexterity. This is in line with the prevalence of additional symptoms affecting the upper extremities in our study.

However, in contrast to other observations (Couch, 1976) we found no correlations between multiple dystonic symptoms and severity as measured by the TSUI-index. The TSUI-index in patients with multiple dystonic symptoms did not differ significantly from the rest of the group. As discussed in a later paper in this series (Scheidt *et al.*, 1996), however, patients with multiple dystonic symptoms tended to be more disabled in their psychosocial adjustment.

The course of ST is usually chronic but intermittent periods of remission can occur. Of the patients 19.1% reported a period of complete remission. This corresponds to other reports (Jayne *et al.*, 1984). Those patients who had experienced a remission were significantly younger than the rest of the group. No correlations were found between course (remission versus non-remission) and severity.

The correlations between the severity of the signs and the neurological symptoms were surprisingly weak. In the later papers in this series (Nickel *et al.*, 1996; Scheidt *et al.*, 1996) what factors other than the neurological signs determine the degree of subjective distress due to physical impairment will be discussed.

Like others, we failed to define subgroups of ST patients except for those clinical forms already established on the basis of the major deviant head position (Lücking, 1980; Deuschl *et al.*, 1992). Multivariate analyses of the data suggests clusters based on more complex groups of variables (Scheidt *et al.*, 1995).

With regard to a genetic predisposition, 3.1% of the patients in this study had a first or second degree relative with ST. This corresponds to the findings of Patterson and Little (1943) (3.8% with a positive family history; $n = 103$) and Rondot (1991) (4.6% with a positive family history; $n = 220$). When other extrapyramidal motor symptoms, such as tremor, and Parkinson's disease were included, 7.2% of our patients had a positive family history. Other studies reported a positive family history of extrapyramidal

motor diseases in 29.4% (van Hoof *et al.*, 1987) and 40.6% (Naber *et al.*, 1986). This difference may be due to the small number of patients enrolled in these studies. Given the low incidence of ST in the general population, the positive family history probably reflects a genetic predisposition, a conclusion in line with other epidemiological studies (Waddy *et al.*, 1991).

In recent papers trauma of the neck has been suggested as a cause of ST (Sheehy and Marsden, 1980; Koller *et al.*, 1989; Jankovic and van Linden, 1988; Truong *et al.*, 1991; Krauss *et al.*, 1992; Goldman and Ahlskog, 1993). In our patients 3.5% had had head trauma within 14 days of onset of the illness. In 9.3% a traumatic origin seemed possible from the history, but could not be proven. These results are comparable to those of Sheehy and Marsden (1980) and Chan *et al.* (1991) both reporting traumatic events preceding the onset of ST in 9%. It seems that trauma should be considered as a cofactor in the aetiology of ST.

No statistical differences were found between the groups "idiopathic torticollis" and "torticollis with positive family history of extrapyramidal motor diseases" in signs, symptoms, severity, duration, age at onset and therapeutic response to treatment with botulinum toxin. The effect of alcohol on dystonic features (Bihary and Koller, 1985) also failed to detect subgroups.

Treatment with botulinum toxin is clearly perceived by the patients as the most efficient form of treatment. Systemic drug treatment caused improvement for a smaller group of approximately 30%. Among surgical procedures stereotactic surgery was of some help in 47%, but this was offset by a worsening in 27%. Physical therapy, a common therapeutic attempt, was experienced as helpful in 34%.

For botulinum toxin we found a response rate of 83.2% without serious side-effects. This result is outstanding compared with other therapeutic attempts and confirmed the effectiveness, availability and safety reported in other studies (Tsui *et al.*, 1986, 1988; Jankovic and Brin, 1991; Poewe *et al.*, 1992; Greene, 1994; Poewe and Wissel, 1994). The successful treatment of ST results in improved professional and social performance (Jahanshahi and Marsden, 1992). However as will be illustrated in the following paper (Nickel *et al.*, 1996) severe disability due to ST persists in different areas of life even under treatment with botulinum toxin.

Acknowledgements

We would like to thank W. Vasold and R. Roos for preparation of the manuscript.

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