

Idiopathic Spasmodic Torticollis: Pathophysiology and Treatment

S. Gauthier

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Idiopathic spasmodic torticollis (IST) is the most common type of primary dystonia encountered in clinical neurology. Although not life threatening, it adversely affects the work ability and social life of those afflicted. This review will summarize the information currently available on the pathophysiology and treatment of IST and its relationship to other dystonic states. Special emphasis will be placed on articles published since the review on spasmodic torticollis by S. Lal in this Journal in 1979.¹

CLINICAL DEFINITION OF IST

We shall define IST as an acquired neurologic syndrome of unknown etiology consisting of involuntary contractions of neck muscles leading to sustained head deviation (Table 1). Such a clinical operational definition is currently required because of the lack of specific structural, neurophysiological or biochemical markers for IST.

IST falls into the broad category of dystonic states as a type of focal dystonia (dystonic movements in a single body part — Fahn, 1984).² If involvement spreads beyond the shoulders, for example to an arm, it would be a type of segmental dystonia (dystonic movements in contiguous body parts). IST may be associated with other forms of focal dystonia such as blepharospasm, writer's cramp, spasmodic dysphonia or orobuccal dystonia. Some patients with IST present a tonic head deviation, some clonic head movements, and some a combination of both. The head deviation can be further divided into torsion and lateral tilt. These patterns are not fixed in an individual patient and can change over time.

The exact incidence of IST is not known, although a figure of 3 per 10,000 is often quoted from unknown sources. Canadian figures for generalized dystonia have recently become available for the provinces of British Columbia and Alberta, giving a prevalence of 4.2 per million.³

IST is certainly the most common type of dystonia seen in Movement Disorders clinics, followed by blepharospasm and the various segmental, hemi- and generalized dystonic states (Table 2). A female preponderance for IST and blepharospasm has been described.^{4,5} As Stejskal and Tomanek⁶ have reported, there is no difference in hand laterality for IST as compared to the general population (90% are right handed). The predominant torsion of the head to the left in their study as well as our patients does not correlate with the dominant hand. The older age of onset of blepharospasm as compared to the other types of dystonia is striking. Only 3 of our 56 IST cases were familial. None progressed beyond neck involvement. The average duration of symptoms was nine years (range of 1 to 64).

LABORATORY FEATURES

We found uninfused brain CT scans to be consistently normal in IST (Lal and Gauthier, unpublished data), whereas one previous study using pneumoencephalography suggested some cortical atrophy.⁷ Positron emission tomography (PET) studies with 2-deoxy-D-glucose have so far failed to show significant abnormalities in IST⁸ whereas at least one patient with hemidystonia has shown decreased oxygen extraction and metabolism,

Table 1: Operational Definition of Idiopathic Spasmodic Torticollis

- acquired, non traumatic
- childhood or adult onset
- clonic and/or tonic involuntary contractions of multiple neck muscles
- sustained head torsion and/or tilt
- for at least 6 months
- often associated with postural limb tremor
- no history of chronic neuroleptic treatment
- no associated ataxia, weakness, spasticity or reflex changes
- normal brain CT scan

and increased blood volume and flow in the contralateral basal ganglia using PET.⁹

Electromyographic studies in IST have shown simultaneous and sustained contractions of antagonist muscles on both sides of the neck during voluntary movements, often associated with various patterns of rhythmic activity.¹⁰ These results are similar to those found in idiopathic torsion dystonia.¹¹

PATHOPHYSIOLOGY

The most promising new data on the anatomical substrate of IST comes from the observation that electrolytic lesions of the interstitial nucleus of Cajal cause frontal torticollis (head tilt) in monkeys and cats.¹² On the other hand, various types of interference (electrolytic lesions or neurotoxin injections) with the nigrostriatal pathway cause horizontal head asymmetry in cats.¹³ Furthermore, neurotoxin injections in the lateral hypothalamus or the locus coeruleus of marmosets causes marked head rotation.¹⁴ Thus, it appears that unilateral lesions in the mesencephalon and diencephalon can cause frontal and/or horizontal head deviation. The clonic component of IST has not been reproduced in animal models as yet.

A form of idiopathic torticollis in ducks has been reported.¹⁵ This included pathological studies of the neck muscles but unfortunately the nervous system was not examined pathologically.

There is no new published information on human post mortem biochemical analysis of the type currently being done in Parkinson's disease.¹⁶ Families and patients suffering from IST will thus have to be made aware of the existence of brain banks.

Since there is usually no progression over time in the involvement of other trunk and limb muscle groups in IST, it is thus unclear how much the current opinion that generalized⁴ or hemi-dystonias^{17,18} originate from a release of premotor cortical activity from thalamic control applies to IST.

TREATMENT

Symptomatic relief for IST can be partially achieved with various medical and surgical treatments. The most commonly used drugs in IST are the benzodiazepines (diazepam, lorazepam) and the anticholinergics (bentropine, trihexyphenidyl). If action tremor of the limbs or prominent clonic activity of the head is present, a beta adrenergic blocker may prove useful. If these drugs fail, various other drugs may help occasional patients.¹ Among the newest drugs, tizanidine was not effective in one study.¹⁹ There are anecdotal reports on the value of marijuana in IST and cannabinoids still remain to be used in well controlled clinical trials. The newest medical therapy consists of

local intramuscular botulinum toxin injections, based on the favorable early results in idiopathic blepharospasm²⁰ and in hemifacial spasm.²¹ The results in IST are encouraging so far.²²

The surgical management of IST varies from sternocleidomastoid muscle excision to peripheral accessory nerve section, bilateral anterior or posterior cervical rhizotomies, or thalamotomy. Our experience at the Montreal Neurological Institute is that while some patients prefer a local accessory nerve section to the more invasive rhizotomies, the latter give a favorable and lasting improvement of head posture in more than half of the patients (G. Bertrand, personal communication).

Nuchal pain from the chronic head torsion or tilt may become a separate problem that requires specific management with physiotherapy, nerve or facette blocks and psychotherapy. Supportive psychotherapy may also be required because of the common depression that follows a change in body image.

The patients also feel socially isolated because of their bizarre head posture. Organizations such as The Dystonia Medical Research Foundation and The National Spasmodic Torticollis Association can provide information and assistance to patients and their families. The usual lack of progression to other parts of the body should be stressed, as well as the possibility (although small) of spontaneous remission.

CONCLUSION

Although there have been no breakthroughs in IST research, there is now an awareness of the number of patients suffering from IST and a hint at the anatomical substrate. Methodology such as whole brain autoradiography in post mortem brains if applied to IST could advance our understanding of the neurotransmitter interactions in dystonia. In vivo receptor studies by PET could also provide useful basic information provided that image resolution is improved. Magnetic resonance imaging may also help in defining subtle structural abnormalities (although head involuntary movements will cause technical difficulties) and hopefully soon spectroscopic nuclear magnetic resonance studies may allow in vivo assessments of basal ganglia activity.

On the treatment side, botulinum toxin injections, if proven to be safe and effective, may provide symptomatic relief at low cost and minimal side-effects.

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Table 2: Dystonic States Treated in 1980-1985 at the MNH Movement Disorder Clinic

| | N | Male/Female | Right/Left/Ambidextrous | Age of Onset |
|---|------------|-------------|-------------------------|--------------|
| Idiopathic Spasmodic Torticollis | 56 (42.4%) | 23/33 | 50/3/3 | 37.6 (11-60) |
| Blepharospasm ± Oramandibular Dyskinesias | 41 (31.1%) | 14/27 | 38/3/0 | 56.4 (30-73) |
| Segmental, Hemi- and Generalized Dystonia | 32 (24.2%) | 16/16 | 26/5/1 | 31.7 (1-54) |
| Writer's Cramp | 3 (2.3%) | 2/1 | 2/1/0 | 34 (30-42) |

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