Role of the vestibular system in the pathophysiology of spasmodic torticollis

Idiopathic spasmodic torticollis is a form of focal dystonia affecting the neck. It is characterised by involuntary repetitive or sustained neck muscle contractions leading to abnormal head postures and impaired control of head movement. With an estimated prevalence of 9/100 000, spasmodic torticollis is the most common focal dystonia. Onset peaks around the age of 40 and there is a female preponderance with a sex ratio of about 1.5:1. Symptoms typically start gradually over a period of days or weeks and remain largely confined to the neck although spread of dystonia to the face (blepharospasm, oromandibular dystonia), larynx (laryngeal dystonia), and arms (dystonic arm postures and writer’s cramp), but not the legs, can occur. Abnormal head and neck postures in spasmodic torticollis cause considerable disability, often interfering with the patient’s ability to work, and cause pain, social stigmatisation, and depression. Most cases are sporadic. However, a family history is reported in about 15% of patients and several families with more than four affected members have been described, all of whom showed autosomal dominant inheritance. Thus genetic factors seem to have some aetiological importance.

Role of the basal ganglia: the double lesion model

The pathophysiology of spasmodic torticollis is still unclear. In idiopathic spasmodic torticollis, no structural lesions are found in the CNS. However, there are two lines of evidence that dysfunction of the basal ganglia plays a part. Firstly, spasmodic torticollis can be seen either in isolation or as part of a more widespread hemidystonia after lesions of the basal ganglia or its connections. Secondly, it is sometimes part of the clinical presentation in generalised primary torsion dystonia, in which functional imaging data and neurophysiological studies have shown abnormalities in the basal ganglia and its projections. If focal dystonias, including spasmodic torticollis, were solely caused by dysfunction of the basal ganglia then the question arises as to why different body parts are predominantly affected in different patients. It has been proposed that there is a rough somatotopy within the sensorimotor region of the basal ganglia, so that spatially limited damage might produce focal symptoms. However, recent functional imaging data have demonstrated considerable spatial overlap between the representation of distinct body segments in the lenticular nucleus, arguing against a clear cut somatotopic organisation of the basal ganglia.

On the other hand, it is possible that the damage to the basal ganglia is more widespread but clinically focal symptoms develop because of a secondary dysfunction in another part of the nervous system. For instance, in a recent animal model it has been shown that blepharospasm can be produced by the combination of a basal ganglia lesion (dopamine depletion in the substantia nigra pars compacta) and a peripheral lesion of the facial nerve, but by neither of these lesions in isolation. Similarly, symptoms may focus on the neck if a subclinical basal ganglia dysfunction, producing widespread abnormalities in the nervous system, is coupled with a lesion affecting any of the systems important for the control of head and neck movement. The existence of widespread abnormalities in spasmodic torticollis has been documented by, for example, the presence of abnormal forearm reciprocal inhibition or blink reflex recovery. Candidates for a focal or segmental lesion would be visual, somatosensory, or proprioceptive pathways from the neck or the vestibular system.

Contribution of abnormal neck afferents

There is no evidence that visual pathways are affected in patients with spasmodic torticollis. On the other hand, abnormalities in somatosensory processing have been described in patients with dystonia. Whereas patients with writer’s cramp were recently shown to have deficits in discrimination of two closely delivered electrical stimuli, to our knowledge there is no study on abnormal somatosensory processing affecting the neck in spasmodic torticollis. However, by analogy with patients with writer’s cramp, whose symptoms have been reported to improve after blocking muscle afferents in the arm, Kaji and Kubori recently showed that muscle afferent block in the neck can also be useful for treating spasmodic torticollis. This implies that neck muscle spindles may be pathologically active in the disorder. The finding of Leis et al of a reduction of involuntary muscle contraction in two patients after neck muscle vibration, which affects spindle discharge, can be interpreted in a similar way. Karnath et al recently reported on a patient with spasmodic torticollis whose symptoms improved after prolonged (15 minutes), but not short, vibration of the dystonic splenius capitis muscle. The authors hypothesised that the delayed improvement in head posture could indicate abnormal central processing of afferent information rather than a peripheral disturbance of proprioceptive afferents.

On the other hand, there is experimental evidence that proprioceptive information from the neck is correctly processed in spasmodic torticollis. In two studies where subjects had to judge the visual vertical and visual straight ahead,
patients with spasmodic torticollis took their trunk midsagittal line as reference, contrary to healthy controls, who relied on the midsagittal plane of their head.\textsuperscript{45} This shift of reference from head to trunk in patients with spasmodic torticollis is perhaps not surprising as an abnormally held, involuntarily moving neck is an unreliable reference. The fact that patients were able to adjust their visual straight ahead correctly to the trunk midsagittal, even during experimental trunk on head movements in the dark (that is, with no vestibular or visual stimulation),\textsuperscript{45} implied that proprioceptive signals coming from the neck were used and interpreted correctly. Also, a study of the cervico-ocular reflex—that is, eye movement responses to rotation of the trunk about the earth fixed head—indicated normal proprioceptive input in patients with spasmodic torticollis.\textsuperscript{46} Therefore, some experiments suggest that somatosensory processing in cerebral and other dystonias is abnormal\textsuperscript{27–29} but not all data support this view.\textsuperscript{30–34} It also remains to be established whether proprioceptive abnormalities are a primary problem driving abnormal neck muscle contractions or a consequence of an imbalance in other systems controlling neck movements—for example, the vestibular system.

**Imbalance and impairment of vestibular reflexes**

Imbalance in the vestibular system and its connections can produce abnormal head and neck postures and movements under experimental conditions. Electrical stimulation of the interstitial nucleus of Cajal, which is closely connected to the vestibular nuclei\textsuperscript{35} and has inputs to the sternocleidomastoid motor neurons in the brain stem and neck motor neurons in the cervical cord,\textsuperscript{36,37} can produce head rotation to the side of the stimulus in cats.\textsuperscript{38} Electrolytic lesions and kainic acid injections into the interstitial nucleus of Cajal in monkeys and cats were associated with contralateral head tilt and intermittent contractions of neck muscles causing “spasmodic” head movements.\textsuperscript{40}

Mild deficits in vestibular reflexes have been described in patients with spasmodic torticollis. The vestibulo-ocular reflex often shows a directional preponderance with the slow phase of the nystagmus induced by rotational and cyclic stimuli being more active ipsilaterally to the head rotation,\textsuperscript{41–43} although some authors have not confirmed this.\textsuperscript{44} These reflex abnormalities persist when the head position is normalised after botulinum toxin injection treatment and cannot be imitated in healthy subjects by forced head rotation,\textsuperscript{45} indicating that they are not merely a consequence of an abnormal head posture. Additionally, reflexive ocular counterrolling during whole body rotation about the naso-occipital axis is pathological in patients with spasmodic torticollis.\textsuperscript{46}

However, other vestibular reflexes are seemingly unimpaired or only mildly abnormal, possibly as a consequence of normal head posture. For example, latency and amplitude of the short latency vestibulo-colic reflex, elicited by sudden, brief hyperextension of the neck using the “head drop” technique\textsuperscript{47} was recently shown to be unimpaired in patients with spasmodic torticollis. Click evoked myogenic responses in the sternocleidomastoid muscle (click evoked vestibulo-colic reflex) were suppressed ipsilaterally to the direction of head turning but this seemed to be related to the duration of the torticollis rather than being a primary abnormality.

**Disruption of vestibulo-voluntary interaction**

Although the short latency (20 ms) vestibulo-colic reflex to a “head drop” is normal in spasmodic torticollis, indicating intact oligosynaptic projections from the labyrinth to the sternocleidomastoid motor neurons, the interaction of vestibular signals with a rapid voluntary neck movement (neck flexion) is not normal.\textsuperscript{48} Münchau et al\textsuperscript{49} studied neck flexion in patients with spasmodic torticollis in three reaction time tasks with the subjects lying supine on a platform: rapid voluntary neck flexion to a simple auditory “go” signal, to a startling stimulus (acoustic startle) and to a strong vestibular stimulus (sudden drop of the head). Voluntary neck flexion responses to an auditory “go” signal and an acoustic startle were normal in patients with spasmodic torticollis. As in normal subjects onset latencies of neck flexion were shorter in the startle reaction time task. In normal subjects onset of voluntary neck flexion in the drop experiment was also significantly earlier than in the simple reaction time task, indicating that, similar to the startle,\textsuperscript{40,41} a vestibular input can speed up or “release” a prepared motor response. By contrast, voluntary responses to the vestibular stimulus in the drop experiment were significantly delayed in patients with spasmodic torticollis when compared with normal controls. In fact, onset of neck flexion in the vestibular drop experiment occurred later than in a simple voluntary reaction time task.\textsuperscript{42} Thus, although an accurate vestibular signal was available to produce a normal short latency vestibulo-colic reflex, it did not interact with higher order level motor processes. This abnormality was unrelated to duration of symptoms, severity, or direction of torticollis, indicating that it is a primary abnormality rather than secondary to abnormal head posture.

As expected, patients with bilateral labyrinthine lesions showed delayed responses to both the short latency response to the head drop and the voluntary reaction to the head drop.\textsuperscript{43–45} Thus, although short latency vestibulo-colic reflex pathways are normal in patients with spasmodic torticollis, their voluntary response after the head drop resembles that of labyrinthine defective patients, as if vestibular information was ignored or not fully integrated at higher CNS levels.

**Abnormal higher order vestibular processing**

A study on head and body posture in patients with spasmodic torticollis also suggested abnormal processing of vestibular information.\textsuperscript{50} The experimental paradigm used neck vibration while standing on a stationary platform. Stationary normal subjects sway forwards on a platform when dorsal neck muscles are vibrated. This is explained as follows: vibration stimulates neck muscle spindles signalling muscle lengthening which, in the absence of corroborative signals from the labyrinth indicating head movement, is interpreted as a backward tilt of the body.\textsuperscript{51} By contrast, patients with spasmodic torticollis do not sway forwards. In them the head deviates backwards during vibration as if the information from the labyrinth, indicating a stationary head, was not taken into account.\textsuperscript{52} In this respect, their response is also similar to that in bilateral labyrinthine defective patients.\textsuperscript{52} Importantly, postural responses to vibration of the Achilles tendon are normal in patients with spasmodic torticollis.\textsuperscript{53} During vibration of the Achilles tendon, both patients with spasmodic torticollis and normal subjects alike sway backwards because vibration is interpreted as lengthening of the gastrocnemius muscle, in turn centrally interpreted as forward sway of the body. In this setting vestibular signals, again signalling a stationary head, are normally “overwritten” and proprioceptive information from the ankles prevails. Thus, context dependent postural responses to vibration are normal in patients with spasmodic torticollis when postural control relies more on proprioceptive information (Achilles tendon vibration) but abnormal when postural control relies on vestibular signals (neck muscle vibration).
vibration). This supports the view that patients with spasmodic torticollis have difficulty with the central integration of vestibular information.

The reasons for abnormal vestibulo-voluntary interaction and higher order vestibular processing are not clear but it can be postulated that the gain, or central weighting, of vestibular input to higher order systems controlling the neck is reduced. Two types of explanation have been put forward why this might happen. Both are based on the assumption that vestibular signals in spasmatic torticollis are in conflict with other sensory signals important for postural control (for example, proprioception), prompting the CNS to down regulate these “unreliable” vestibular signals. Firstly, such down regulation might occur because involuntary head movements lead to constant stimulation of the labyrinth out of context of whole body posture. This explanation implies that abnormal vestibular processing is secondary to abnormal head movements. Secondly, subtle vestibular abnormalities in spasmatic torticollis may pre-exist independently of abnormal head movements. Some of these abnormalities could arise from structures particularly involved in the control of head posture (for example, the extrathalamic nucleus of Cajal) and this would explain the general concordance between the direction of the torticollis and the bias in the vestibulo-ocular system.

Coupling of abnormal vestibular processing with basal ganglia deficits

Under normal circumstances, such vestibular deficits may produce no longstanding head postural problems, as is exemplified by the numerous patients with vestibular lesions (peripheral or central) who do not develop torticollis. However, by analogy with the animal model of experimental blepharospasm, when subtle vestibular deficits are coupled with basal ganglia abnormalities, compensatory mechanism may fail and trigger the development of torticollis. In this respect two findings are of particular interest.

Firstly, there are reports of patients who developed spasmatic torticollis after eighth nerve or vestibular lesions. Unilateral eighth nerve lesions lead to a vestibular imbalance but in otherwise normal patients this is not sufficient to produce torticollis or to cause abnormal sternocleidomastoid activity during backwards tilt. Apparently, in otherwise normal patients biased vestibular output is compensated by central modulation. However, in a study of three patients who developed spasmatic torticollis after an eighth nerve lesion, neck muscle EMG was enhanced during backwards tilt implying diminished central modulation of asymmetric peripheral vestibular input. Conceivably, spasmatic torticollis in these patients was the result of the combination of an eighth nerve lesion producing a vestibular output bias and impaired control of brain stem excitability due to pre-existing altered basal ganglia function.

Secondly, in about 10% of patients with spasmatic torticollis there is a history of head and neck trauma, albeit mild in most cases, before the onset of symptoms. The vestibular apparatus is extremely sensitive to trauma. Such a trauma may cause subtle subclinical vestibular abnormalities that would become symptomatic only in susceptible people.

Consequences for postural control of head and body

If vestibular processing is disturbed in spasmatic torticollis this should have functional consequences in whole body postural control. Indeed, whereas patients with spasmatic torticollis are as stable as healthy controls on conventional (static) force platforms, they show increased body sway when standing on a moving platform. Assuming that these findings genuinely reflect decreased postural performance (for example, the increased whole body sway could just be due to increased neck dystonic activity during the dynamic task) they could be interpreted as follows. The abnormal vestibular input in spasmatic torticollis has no effect during static balance because other sources of afferent input can be used to stabilise posture. However, during the more difficult task (moving platform), all sources of sensory input become important, and therefore vestibular deficits become functionally relevant. In line with this view is the finding that abnormalities of voluntary modulation of the vestibuloocular reflex in the “head drop” study were correlated with the patients’ motion-dependency of neck symptoms. Patients with more abnormal higher order vestibular processing (assessed by the delayed onset of neck flexion EMG activity in the active head drop experiment) had more difficulties in controlling head and neck movements during daily life—for example, during sudden jolts while travelling in vehicles.

How could the concept of abnormal higher order vestibular processing in spasmatic torticollis be reconciled with one of the most characteristic features of spasmatic torticollis, the gesang antagoniste—that is, the use of sensory tricks by the patient to normalise head posture? These tricks include altering cutaneous input (for example, touch of the face or neck) or performing stereotyped movements (for example, lifting one arm and approaching but not necessarily touching the face) thereby inducing proprioceptive or efferent copy signals. It could be speculated that in a situation where one important system controlling head and neck movements is deficient, the nervous system can compensate by relying on additional sensory cues. The local neck response to posterior neck muscle vibration in patients with spasmatic torticollis might be interpreted in the same way, in the sense that such patients show an increased responsiveness to somatosensory stimulation. Alternatively the gesang may indicate that sensory dysfunction in spasmatic torticollis is wider involving vestibular as well as additional sensory inputs.

Conclusion

There is little doubt that abnormalities in the basal ganglia are involved in the pathophysiology of spasmatic torticollis. On the other hand, some vestibular abnormalities have also been reported that cannot solely be explained as being secondary to abnormal head and neck movements in spasmatic torticollis but seem to be more intimately related to its pathophysiology. In this respect, abnormal interaction of vestibular signals with higher order motor commands and disrupted central vestibular processing, which are perhaps caused by subtle imbalances in the vestibular system, seem to be of particular importance. Although a possible vestibular contribution to the origin of spasmatic torticollis remains speculative, the current view would be that spasmatic torticollis might develop when there is a combination of a propensity towards dystonia, produced by a basal ganglia deficit, coupled with vestibular deficits. This is perhaps best illustrated by reported patients who developed spasmatic torticollis after eighth nerve lesions, presumably because of pre-existing subclinical basal ganglia abnormalities. However, other systems important for head and neck movement control, especially proprioception, may be additionally affected and contribute to the disorder.
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