

**Blepharospasm-oromandibular dystonia syndrom
(Brueghel's syndrome)
A variant of adult-onset torsion dystonia ?
C. D. MARSDEN**

From the University Department of Neurology, Institute of Psychiatry, and King's College Hospital School, Denmark Hill, London

Idiopathic blepharospasm is well recognised, but idiopathic oromandibular dystonia is not. The latter consists of prolonged spasms of contraction of the muscles of the mouth and jaw. These dystonic movements appear to be distinct from the chewing, lip smacking, and tongue rolling choreiform movements that characterise the more familiar orofacial dyskinesia. Orofacial dyskinesia, whether drug-induced or spontaneous in origin, is not discussed further in this paper.

Both blepharospasm and oromandibular dystonia may occur in patients with known diseases of the basal ganglia such as those that cause symptomatic torsion dystonia, or may be produced by neuroleptic drugs. They may present also as isolated dyskinesias in middle or late adult life in otherwise normal people. Both have been regarded by many as 'psychogenic'. However, I have now seen 39 patients with these conditions, and the data they afford lead to the conclusion that they are diseases which may be focal manifestations of the syndrome of adult-onset torsion dystonia.

Idiopathic blepharospasm and oromandibular dystonia may occur separately or together. Hence the suggested title 'blepharospasm-oromandibular dystonia syndrome', which may have been described first by Meige (1910). However, I am grateful to Dr R. E. Kelly for pointing out that Pieter Brueghel The Elder, clearly recognised the syndrome. More recently Altrocchi (1972) described two patients with isolated oromandibular dystonia, and Paulson (1972) reported three patients with blepharospasm and oromandibular dystonia.

Case material

All the patients to be described were in normal health at the onset of their illness, were taking no drug known to provoke dyskinesias at that time, and had no other neurological or mental deficit. I have seen 13 patients with isolated idiopathic blepharospasm, 17 with both blepharospasm and oromandibular dystonia, and nine with isolated idiopathic oromandibular dystonia. In all three conditions, women were more frequently affected than men (25 females, 14 males); the peak age of onset was in the sixth decade; and so far the illness had persisted on average for five years. None gave a family history of a similar illness.

BLEPHAROSPASM ALONE The 13 patients with blepharospasm all presented with spasms of contraction of orbicularis oculi causing eye closure. Such spasms sometimes started in one eye only, but both eyes were affected eventually. Typically the spasms occurred only occasionally to begin with but became more frequent and prolonged with time. They might last from a few seconds to as long as 20 minutes or so, and could occur as frequently as every 15 to 20 seconds. Many of these patients were rendered functionally blind, and could not leave the house or cross the road. The spasms were provoked characteristically by bright sunlight (many wore dark glasses), watching television or reading, embarrassment, and fatigue. Many patients had discovered tricks to open the

eyes, including forced jaw opening, yawning, neck extension, or prising them open with the fingers. Many developed a secondary blepharitis due to the spasms. Power of eye closure was normal in all, as were lid and external ocular movements.

Six patients were depressed before and at the onset of their blepharospasm, but seven were in normal psychiatric health. Two of the latter had local eye conditions at the onset, a conjunctivitis in one and a Meibomian cyst in the other, but the remainder had no physical illness.

BLEPHAROSPASM WITH OROMANDIBULAR DYSTONIA Blepharospasm and oromandibular dystonia coexisted in 17 patients. In 13 the two dyskinesias appeared at the same time; in four blepharospasm antedated oromandibular dystonia by one to three years. The blepharospasm was identical with that occurring in the first group of patients described with that dyskinesia alone.

The oromandibular dystonia varied in its clinical manifestations, but in all cases consisted of spasms of the muscles of the face and jaw, and sometimes the tongue. The spasms lasted for up to a minute, and were often repetitive but irregular in timing. They could occur spontaneously, but often appeared on talking or chewing, at least initially. The jaw might be forced open and the lips retracted with spasm of platysma, and the tongue protruded. One patient suffered recurrent dislocation of the jaw, and in another the spasms of jaw opening were strong enough repeatedly to snap misapplied dental wiring. In other patients, the spasms would forcibly close the jaw and the lips would purse like a fish (Fig. 2). Such spasmodic jaw closure caused laceration of the lips and tongue and spasms of the jaw, mouth, and tongue led to inability to wear dentures, chew or even to speak clearly. Thirteen of the 17 patients had difficulty in speaking, while five had difficulty in swallowing. Voluntary movements of the face, jaw, tongue, and palate were of normal strength and rapidity, but such attempted movements frequently provoked muscular spasms.

Five of the 17 patients were depressed before and at the onset of the illness, but the other 12 were in normal psychiatric health. Three similar patients have been described by Paulson (1972).

OROMANDIBULAR DYSTONIA ALONE Nine patients had isolated oromandibular dystonia, with spasms of mouth and jaw closure in five and of mouth and jaw opening in four. Contractions of the lower perioral facial muscles and platysma accompanied the jaw spasms in all patients, but the eyes were spared. The oromandibular dystonia in these patients was similar to that seen in those with blepharospasm as well. In seven patients the jaw would open forcibly with mouth retraction, and sometimes tongue protrusion (Fig. 3). In the remaining two patients the spasms were of jaw closure and mouth pursing. Speech was affected in all, and the initial difficulty was confined to muscle spasms on speaking in four patients and on singing in one. One patient could not speak without developing spasms of jaw opening but could sing normally. Another initially developed spasms of jaw clenching on singing high notes but could speak normally. Both subsequently developed spontaneous muscle spasms of the jaw and mouth. All had difficulty in eating and four had difficulty in swallowing. Three were depressed before and at the onset of their illness, but six were in normal psychiatric health. Two similar patients have been described by Altrocchi (1972).

COURSE OF ILLNESS The duration of the illness varied from one to 28 years. None of the patients with isolated blepharospasm developed dystonia elsewhere. Two of the patients with oromandibular dystonia but not blepharospasm presented initially with dystonia elsewhere. In one, the initial complaint was of retrocollis, followed by retraction of the lips and jaw clenching three years later; in the other, a dystonic writer's cramp of both arms was followed by spasms of jaw opening and inspiration some 10 years later. The other seven patients with oromandibular dystonia

without blepharospasm had no dystonia elsewhere.

Eight of the 17 patients with both blepharospasm and oromandibular dystonia developed dystonia elsewhere. In three of these patients spasms of the respiratory muscles occurred within one to seven years of the onset; these usually consisted of spasms of inspiration accompanying jaw opening or closure and eye closure. One man developed torticollis with retrocollis and spasms of the respiratory muscles some 18 months after the onset of the blepharospasm and oromandibular dystonia. Two women developed antecollis and another developed flexion spasms of the trunk within three years of the onset. Another woman developed torticollis with the chin turned to the left, and tremor and dystonic posturing of the left arm 18 months after the onset. None of the 39 patients developed intellectual, pyramidal, cerebellar, or sensory deficits. Nor did they exhibit akinesia or rigidity (in the absence of drugs). None had fits.

INVESTIGATIONS No consistent abnormality was found in these patients on investigation. Skull radiographs, brain scan, electroencephalogram, cerebrospinal fluid examination, serology, plasma calcium and copper studies were normal when undertaken.

TREATMENT Many therapeutic avenues have been explored. None cured the illness, and no spontaneous remissions were observed. The following groups of drugs have been tried: benzodiazepines (including clonazepam), tri-cyclic antidepressants, monoamine oxidase inhibitors, anticholinergics, amantadine, levodopa, dexamphetamine, tetrabenazine, reserpine, phenothiazines, butyrophenones, pimozide, carbamazepine, barbiturates, tryptophan, and 5-hydroxytryptophan with carbidopa. Most were quite ineffective. Anticholinergics and benzodiazepines gave a little relief, while drugs acting by dopamine receptor blockade (reserpine, tetrabenazine, phenothiazines, butyrophenones, and pimozide) sometimes helped the oromandibular dystonia but not the blepharospasm, often at the expense of unacceptable drug-induced Parkinsonism. Successful treatment of any accompanying depressive illness allowed the patients to tolerate their disability more readily but did not cure the movement disorder.

One patient required surgical division of cricopharyngeus to relieve severe dysphagia, and two patients with blepharospasm had undergone surgical section of the upper branches of the facial nerve to paralyse orbicularis oculi, with partial success. None had been submitted to stereotaxic thalamotomy.

Individual patients had undertaken behavioural therapy, aversion therapy, psychotherapy, hypnosis, and acupuncture, but with no cure. As in the case of successful treatment for depression, techniques aimed at adjusting the individual's reaction to the illness allowed it to be borne with less distress.

DISCUSSION I would draw three main conclusions from these data: (1) blepharospasm and oromandibular dystonia are manifestations of a single illness or syndrome; (2) this is a physical illness, not a manifestation of a psychiatric disorder; (3) this syndrome is related to idiopathic torsion dystonia-hence the title 'blepharospasm-oromandibular dystonia syndrome'.

This group of patients represents a consecutive series referred for neurological opinion. The fact that 17 of the 39 cases had both blepharospasm and oromandibular dystonia strongly hints at their common origin. Blepharospasm, or oromandibular dystonia when it occurred alone, was clinically identical with that seen when the two appeared together. The conclusion is that blepharospasm and oromandibular dystonia have a common pathophysiological basis.

To prove rigorously that a physical complaint is due to a psychiatric illness (of mood or thought), it is necessary to show that such an illness is present at the onset of the physical disorder, that it

persists if the physical disorder continues, and that the physical disorder remits if the psychiatric condition is cured. Fourteen of the 39 patients were depressed at the onset of the illness, but 25 were judged not to be so. Others became depressed or anxious because of their physical illness, but at least 10 stalwart and robust individuals withstood their physical disability without mood change. Treatment of any additional depression did not cure the physical illness. On these grounds I would not consider the blepharospasm oromandibular dystonia syndrome as due to psychiatric disability. (However, psychiatric treatment may be necessary and rewarding in an otherwise intractable condition.)

If the syndrome is due to a physical disorder, what are the clues to its origin? Evidence exists to attribute it to a disorder of the basal ganglia. Thus blepharospasm and oromandibular dystonia were caused by encephalitis lethargica, and blepharospasm occurs in Parkinson's disease. I have seen both provoked by neuroleptic drugs, such as phenothiazines or butyrophenones. The typical facial movements of the chronic tardive dyskinesia caused by neuroleptics consist of choreiform chewing, lip smacking, tonguing, and licking (the classical orofacial dyskinesia). However, on occasions I have seen the more prolonged and repetitive spasms of contraction of mouth and jaw muscles that characterise oromandibular dystonia, and also typical blepharospasm, in such drug-treated patients. Drug-induced chronic tardive dyskinesias are believed to be due to the effect of neuroleptics on dopamine receptors in the corpus striatum (cf. Marsden et al., 1975). I have also seen blepharospasm and oromandibular dystonia in some patients with athetoid cerebral palsy and other conditions causing symptomatic torsion dystonia in which the pathophysiological basis of the motor disorder is believed to be lesions in the basal ganglia. The tentative conclusion is that the idiopathic blepharospasm oromandibular dystonia syndrome is due to abnormal function of the basal ganglia or some other extrapyramidal centre.

The characteristics of the muscular spasms in the disorder, their prolonged duration, their repetitive pattern but irregular timing, and their provocation by voluntary action are typical of the abnormal muscular spasms of torsion dystonia. Classical idiopathic torsion dystonia or dystonia musculorum deformans is a disease of childhood and adolescence, often inherited, with onset of dystonic muscular spasms in legs, arms, or neck, and usually progression to generalised involvement with severe disability. A similar illness occurs in adults, but differs in that it is rarely inherited, the arms and axial muscles are almost always first involved, and progression to involve other body parts is limited (Marsden et al., 1976). Blepharospasm-oromandibular dystonia syndrome resembles adult-onset idiopathic torsion dystonia in all these respects, and resembles other focal dystonias which may represent isolated manifestations of adult-onset torsion dystonia, such as spasmodic torticollis and dystonic writer's cramp (cf. Marsden, 1976a, b, for discussion of the evidence to suggest that these entities are focal dystonias). The peak age of onset of adult idiopathic torsion dystonia, torticollis, and dystonic writer's cramp in the fifth decade of life is close to that of blepharospasm-oromandibular dystonia syndrome. Blepharospasm and oromandibular dystonia occurred in 9% and 43% of 47 typical cases of idiopathic torsion dystonia with onset in childhood (Marsden et al., 1976). In the present series of patients, a minority presented with, or subsequently developed, torticollis or dystonic writer's cramp. All the evidence points to the conclusion that blepharospasm oromandibular dystonia syndrome is another manifestation of adult-onset torsion dystonia, and that it is a focal dystonia akin to spasmodic torticollis and dystonic writer's cramp.

As to its fundamental cause or causes, there is no information. No pathological or biochemical examination of the brain in such cases has been reported and there is an urgent need for such studies.

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