

Cranial dystonia

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ABSTRACT

Cranial Dystonia is characterized by three distinct components - blepharospasm and spasm of oromandibular and laryngeal musculature. We report a case of 73-year old male with features of oromandibular and laryngeal dystonia. Clinical symptomatology, clinical investigations including EMG studies and the progress of the case with drug and speech therapies are described.

Keywords: Cranial dystonia; Cervical dystonia.

INTRODUCTION

One of Brueghel's paintings from the 16th century bears a remarkable similarity to what was later described in the clinical features of cranial dystonia by Meige in 1910. Dystonia is a term applied to strong, sustained or intermittent muscular spasms resulting in extreme degree of flexion or extension. There are at least 3 recognizable and distinct components of cranial dystonia viz. blepharospasm, oromandibular and laryngeal. Blepharospasm entails frequent spasmodic eye closures. Oromandibular dystonia affects muscles around the lower face, mouth and tongue causing dysphagia. Laryngeal

dystonia results in forced spasm of laryngeal muscles producing a monotonous high pitched and strained voice. Each of the above may occur in isolation but the more common clinical situation is combination of two components. We present here a case which showed a combination of oromandibular and laryngeal dystonia.¹⁻⁵

CASE REPORT

History

KG, a 73-year male, presented with gradually progressive tightness of jaw and neck muscles with difficulty in speaking for

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one year followed by difficulty in swallowing for 7 months. The latter was more for liquids than solids. At times it was accompanied by vomiting and violent coughing. Of late the patient also complained of excessive salivation while eating or chewing. All the complaints aggravated while chewing or consciously performing any act with the involved muscles. The discomfort/spasm remained the same even when he lay down in supine position. However, on changing side to left or right lateral in lying position, there was appreciable and considerable loosening of the muscles of jaw, neck and mouth. He had no eye complaint in the form of blepharospasm. He lost about 4-5 kg of weight in the past one year. There was no history of unconsciousness, paralytic stroke, epilepsy, head injury, memory disturbances, psychiatric illness or treatment with anti psychotic medication. He reported no tightness in limbs, tremors, difficulty in walking or maintaining balance, decrease in speed of movement, difficulty in initiating movement, disturbances in the special senses. There was no bladder and bowel dysfunction.

His personal history showed that he had joined the British Gurkha as a driver during the 2nd World War and served them for about 17 years which included overseas assignment at Hong Kong. He returned to his native village in the hills in Distt Lamjung, about 10 hrs of trekking from the nearest road head in 1961. For the last 37 years, he has been subsisting on farming and meagre pension from the British Gurkha. He has a wife and 4 children: 3 daughters and one son. His two daughters are married and living happily. His youngest daughter aged 23 is unmarried and living with

him. His son aged 16, is being brought up by his sister-in-law close to Pokhara. His wife is suffering from chronic abdominal problem for which she has not received any proper medical aid. On the whole his psychological space shows areas of distress which are not unduly serious.

Examination

Our patient is a thinly built individual who bears somewhat tense facial expression and speaks in barely audible voice. Phonological examination showed that all sounds were affected except for bilabials eg, p, ph, b, bh. Oral examination showed that the tongue had diminished movements in upward and lateral direction. Close examination of the swallowing showed lack of proper coordination in the swallowing process. He had pronounced difficulty in swallowing liquids in particular. He was found to be well oriented with time, place and person. His ability to recall recent and past events was not affected. His comprehension, insight and judgement remained unimpaired. His emotional reactions during repeated interviews with all three psychiatrists who were involved in this case, were evaluated to be normal and resonant. His Mini Mental Status Examination score was normal with a score of 30.

He had no evidence of hypertension, pallor, oedema, clubbing, jaundice or lymphadenopathy. Facial musculature did not reveal any asymmetry. However, there was discernable increase in muscle tone in the neck muscles (mainly the sternocleidomastoid and platysma), jaw muscles, slight restriction in the side to side movements of the tongue (which was not

deviated) and also pursing movement of the lips.

There was no evidence of any cranial nerve palsy. He had no deviation of mouth, nasal regurgitation, loss of gag or palatal reflexes and taste disturbances. Power and bulk of all muscle groups was normal. Excepting those described above, no rigidity was present in any other muscle groups including respiratory muscles. Tremors or any adventitious movements or gait disturbances suggestive of pyramidal or extra pyramidal dysfunction were not seen. No cerebellar or meningeal signs were elicited. Oto rhino laryngological examination including indirect and fibre optic laryngoscopy showed normal vocal cord movement and no pathology like growth, ulcers or evidences of trauma were observed.

Investigations

Haematological, Hb 12.3 gms%, TLC 8000 cells/Cmm, DLC P50, L39, E1%, ESR 12 mm in 1st hr, Peripheral blood smear showed normocytic, normochromic red blood cells and normal leucocytic morphology.

Biochemistry:

Fasting Blood glucose 80 mg%, Blood Urea 30 mg%, Serum Cholesterol 165 mg%.

CALIBRATION PROTOCOL

CAL: 5 MM = 50 MICROVOLTS

MUSCLE & ACTIVITY

Serology:

VDRL and HIV were negative.

Radiological:

X-ray chest and skull were normal.

CT Scan brain detected no abnormality.

ECG:

Normal

EMG examination:

EMG was taken on a Bio potential Coupler. Electrodes were placed over masseter and laryngeal muscles. The sensitivity of the recorder was kept at 100 microvolts for a deflection of 5 mm and paper speed was kept at 25 mm per sec. The record was carried out during baseline as well as during various acts of bringing the muscles under contraction. It was done for normal control as well as the patient in the same sitting. The analysis of the EMG record (Fig. 1) shows that there was a definite evidence of hyperactivity of the muscles at baseline and the EMG during contractions was either not sustained properly or became too bizarre.



CONTROL

SUBJECT

MASSETER
BASELINE

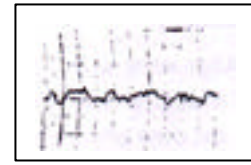
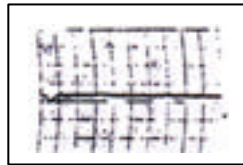
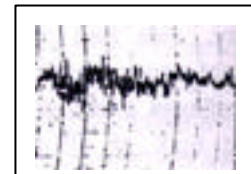
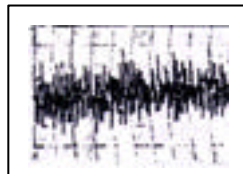
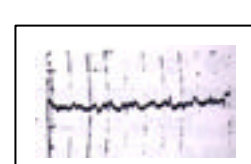
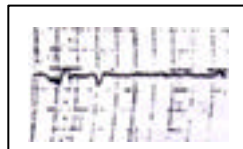


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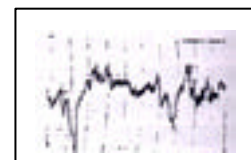
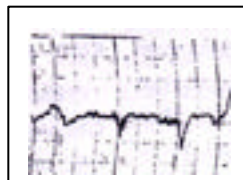
MASSETER
CONTRACTION



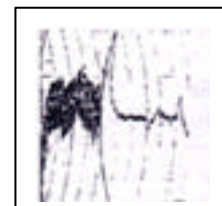
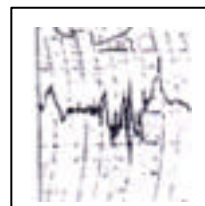
LARYNGEAL
BASELINE



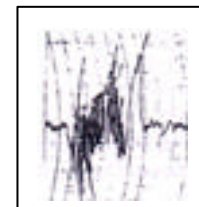
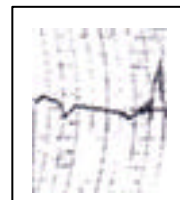
LARYNGEAL
CONTRACTION
(Phonation)



LARYNGEAL
CONTRACTION
(Coughing)



LARYNGEAL
CONTRACTION
(Swallowing)



IMPRESSION: The subject shows much higher baseline potentials in the affected muscle groups compared to the control. The contractions also show higher built up of EMG potential.

Fig. 1: EMG studies: case report

Treatment

After initial work up the patient was put on Trihexyphenidil. He was initially started on 4 mg dose which was increased at weekly or biweekly intervals depending upon improvement in clinical condition and tolerance of side effects. He was on a regimen of 10 mg of Trihexyphenidil by 4th week. It was noticed that any further increase was not likely to be tolerated well. He was also put on intensive speech therapy in a systematic manner. Speech therapy was aimed towards tongue and lip exercise and improved articulation drill. He showed considerable improvement in swallowing of solids/liquids but speech showed only marginal improvement. The chronological progress of the case was diligently observed and noted. This is shown in Table I.

Table I: Chronological progress

<i>Week</i>	<i>General</i>	<i>Speech</i>
1-2	Tonicity of neck muscles ie, sterno cleidomastoid and platysma, decreased	Alveolar plosives showed slight improvement eg. t, th, d, dh
3-4	Tonicity further decreased with improvement in swallowing solid food	Alveolar plosive improved to word level

5-6	Swallowing of solids markedly increased with minimal change in speech. At this dosage, there was no improvement in swallowing liquids which continued to result in coughing or vomiting despite the use of straw or spoon to take in fluids.	Alveolar plosive improved to phrase level.
7-8	Patient was able to swallow small quantity of thick liquid with spoon but difficulty in swallowing thin liquid persisted.	Velar sound eg. k, kh, g, gh slightly improved in isolation.
9-10	Patient was able to swallow well along with consumption of fluids in small gulps.	Velar improved to word level. Alveolar plosive to sentence level and overall speech legibility was only slightly better.

DISCUSSION

Oromandibular dystonia commonly occurs in conjunction with blepharospasm but maybe seen in an isolated form. The clinical picture is variable, ranging from spasm of jaw opening or closure or deviation of the mandible from the mid line. Forward protrusion of the jaw maybe associated with evident contraction of the adjacent neck muscles especially the platysma. Tongue maybe involved concurrently or singly, being protruded

intermittently or in any of direction within the oral cavity with feeding difficulty. In association with jaw spasm or separately lips maybe pursed or drawn inwards. Spasm of the tongue, protrusion or jaw closure may result in self injury, sometimes as severe as tooth damage or gum laceration. In most patients, dystonic spasm are present at rest often exacerbated by any task involving the jaw, face or tongue muscles. Spasm increases on speaking, chewing (action dystonia), embarrassment, or fatigue which is more marked as the disease progresses. As in other dystonias, spasm abates during sleep and patient may develop trick manouevres to gain control over their involuntary movements.³⁻⁴

Laryngeal dystonia results in a high pitched strained voice as if the patient is being strangled. Adduction spasm of the laryngeal muscles on attempted phonation is the usual cause. While speaking, voice maybe severely compromised. Other vocal tasks like whistling or singing may be preserved. The voice may sound normal at the onset of speech becoming subsequently strained. Laryngeal dystonia maybe seen with other focal dystonias generalized dystonias and benign essential tremors.⁵⁻⁶

Cranial dystonia is more common in females, and has a peak incidence in the 5th, 6th or 7th decades of life as against tortion dystonias which is a disease of childhood and adolescents with lower limb dystonia spreading to involve axial, upper limb and finally cranial structures. If the disease

begins in an isolated fashion, it may spread to involve adjacent cranial structures eg, oro mandibular dystonia followed by laryngeal dystonia. The established syndrome of spasm of eye closure and tongue protrusion with or without a strained voice constitutes a typical and easily recognizable clinical picture. Physical examination is unremarkable apart from spasm occasionally torticollis, respiratory gasp or grunts, or tremors of outstretched limbs. Rarely a family history of similar illness maybe obtained. Patients may become depressed during the course of this persistent and disabling disease.⁷⁻⁸

In idiopathic cranial dystonia there is no evidence suggestive of epilepsy, dementia, involvement of motor/sensory/cerebellar pathways. The movement disorder maybe secondary to brain stem vascular disease or demyelinating diseases. Treatment with anti psychotic medication may result in oro facial (Tardive) dyskinesia. Orofacial dystonias consist of repetitive prolonged spasm rather than incessant flow of chorieforn lip smacking, chewing and tongue rolling movement seen in tardive dyskinesias. Neuroacanthocytosis is a rare condition in which patient may present with eating difficulties, as spasms of tongue extrude food from the mouth as soon as the eating begins. An increased number of acanthocytes is seen in wet film preparation of peripheral blood.⁶

The association of blepharospasm with oromandibular dystonia supports the view that blepharospasm is likely to represent some

subtle form of extra pyramidal dysfunction although direct evidence is lacking. There maybe occasional association with parkinson's disease, may show side effects of treatment with anti psychotics or L-dopa; maybe associated with Wilson's disease, torsion dystonias or following encephalitis lethargica is noticed. A close relationship between dystonia and athetosis has been observed. Putamen can be involved in both but in dystonias, thalamus and cerebral cortex maybe involved as well. Neurophysiological abnormality in patients with blepharospasm and oromandibular dystonia maybe related to an abnormal excitatory drive on to brain stem structures mediating such reflexes. Possible psycho genesis has also been considered. Pathology is largely undefined.^{5,6,9}

As pathology of cranial dystonia remains uncertain, treatment is difficult and largely empirical. High doses of anticholinergics such as Bzotropine/Benzhexol/Trihexyphenidil are of considerable and persistent benefit in significant number of both generalized and focal dystonia. Commencing treatment with conventional dose of 4-6 mg per day with gradual increases of about 2 mg per week until side effects occur or benefits are ceased have been suggested. Younger patients tolerate treatment better than older patients. The aged are likely to experience mental confusion or urinary retention which may abort treatment. Neuroleptics are contraindicated.⁹⁻¹¹ The use of Benzodiazepines has also been advocated in the treatment of these disorders.⁹

In addition, surgical procedures like nerve section/avulsion, have been used for blepharospasm as well as laryngeal spasm. Technique of nerve thermolysis has also been developed for orbicularis oculi. However, injection of Botulinum toxin type A is preferred for blepharospasm and laryngeal dystonias. The improvement with these regimens has evoked mixed response. Other treatment modalities like behaviour therapy, hypnosis, acupuncture and psychotherapy have been tried without either consistency or long-term benefits.¹²⁻¹⁴

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