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NEUROACANTHOCYTOSIS : A CASE REPORT

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ABSTRACT

An adult male with severe tic disorder presented with recurrent injury to tongue and dysphagia. There were a significant excess of acanthocytes in the peripheral blood smear. Treatment with lithium resulted in a considerable reduction in the severity of tics and self-injurious behaviour.

Key words : TICS, acanthocytes, self-injury

It is well recognised that neuroleptic induced dyskinetic movements involving the oral, buccal and lingual group of muscles can result in significant dysarthria and dysphagia (Pi Eh and Simpson,1981; Gregory et al.,1992). However, it is uncommon for such patients to injure their lips and tongue. Occurrence of repeated injury to tongue and lips in a patient with oral dyskinetic movements suggests a diagnosis of a non-drug induced progressive neurological disorder such as neuroacanthocytosis (Hardie et al.,1991). In this report we present a case of an adult male with severe tic disorder who also had recurrent injury to tongue in the form of bites and lacerations. This patient had received neuroleptics for many years for the treatment of tics and had also developed drug indued dyskinetic movements.

CASE REPORT

A thirty six year old male was admitted on three different occasions to the Department of Psychiatry, between 1996-1999, with complaints of recurrent tongue bites and difficulty in swallowing. He had multiple lacerated wounds and scars on the lateral edges of the tongue. The patient could prevent and minimise these tongue bites by keeping a piece of cloth in the mouth. He was judged to have normal intellect but was depressed since he had no control over these injuries. He had progressive difficulty in chewing and swallowing food. He also had multiple motor tics in the form of jerky movements of the trunk, shoulder, neck, limbs and vocal tics. According to the patient's mother, tics involving neck and shoulder had started 20 years ago. These movements had gradually increased over the last six years and led to social avoidance, poor work attendance and poor marital relationships. Since 2 years, the patient had been receiving haloperidol for control of tics. At no point of time during the course of the illness did he have any symptoms suggestive of psychosis or dementia. He has had several generalised tonic clonic seizures since 1995 for which he was receiving carbamazepine 600 mg/day.

The proband was the only affected sibling and was second of six children born to consangunious healthy parents. His birth and childhood development were uneventful. He was a bright student and got married after graduating from college. His only son aged 10 years is healthy and free from tics.

Before his admission to our department in 1996, the patient had undergone a series of laboratory investigations including blood tests, brain CT scan and MRI. Laboratory investigations did not reveal any abnormality. A diagnosis of tic disorder with neuroleptic induced dyskinesia was made. He was started on

Clonazepam 0.5 mg, and the dose was gradually increased to 1.5 mg/day. However, clonazepam had to be discontinued due to excessive day time sedation. Subsequently he was started on clonidine and on a dose of 0.4 mg/day, the abnormal movements and injuries to the tongue decreased considerably. The patient was discharged home on clonidine 0.4 mg/day.

The patient was readmitted to our hospital within a month with recurrence of tongue injuries, movement disorder and difficulty in swallowing despite being on regular medication. Physical examination also revealed hyporeflexia, hypotonia in both the lower limbs suggestive of bilateral sensori-motor neuropathy. ENMG showed demyelinating sensori-motor neuropathy. The combination of tics, neuropathy and self-injurious behaviour was suggestive of a diagnosis of neuroacanthocytosis. Peripheral blood smear examination done on many occassions during this admission showed an excess of acanthocytes (31% of acanthocytes) (Table). Clonidine was then discontinued and patient was started on triperidol. On a daily dose of 3.5 mg of triperidol there was a significant improvement in the patient's clinical status. After discharge from the hospital, patient did not come for follow up visits for almost one year. During this time he was treated elsewhere and had received Botulinum toxin to prevent severe recurrent tongue injuries. Triperidol had been discontinued and patient was receiving pimozide 10 mg/day and trihexyphenydyl 8 mg/day.

Patient came back to us in 1999 with reappearance of the tongue bites, tics and dysphagia. Patient was started on tetrabenazine 25 mg/day and the dose was gradually increased to 175 mg/day. However tetrabenzine had to be discontinued as patient developed severe drug induced parkinsonism and hypotension. At this point of time lithium carbonate was introduced. There was a remarkable improvement in the patient's clinical status and he was discharged on 1050 mg of Lithium (plasma level 0.5 mEq/ litre). Neuropsychological testing done during the present admission did not reveal any deficit. Patient is currently ambulatory and is free from tongue injuries and tics. However the symptom of dysphagia persist and and he is receiving nutrition through gastrostomic feeding tube.

DISCUSSION

Acanthocytes are abnormal red blood corpuscles detected in fresh blood films by their spiny protuberences (must be distinguished from echynocytes/artefacts). The pathogenesis in acanthocytosis is due to an abnormal composition of covalently bound fatty acids in erythrocyte membrane protein (Sakai et al.,1991). A spectrum of neurological and behavioural abnormalities viz., depression, anxiety, obsessive compulsive symptoms, personality change and cognitive impairments have been associated with neuro-acanthocytosis (Hardie et al.,1991). The onset and clinical progression of neuroacanthocytosis resembles Tourette's Disorder and Huntington's Disease.

Among the four categories of Neuroacanthocytosis (NA) described (Jankovic and Janet Orman,1988), our case belonged to the group NA with normal lipoproteins (Table). The other categories are : NA with hypobetaproteinemia, NA with abetalipoproteinemia (Bassen-Kornzweig disease) and x-linked NA (McLeod syndrome).

In our patient various dopamine antagonists

TABLE
INVESTIGATIONS

Haemoglobin	13.7 gm	RBC counts 4.76 m/cu mm	
PCV	48.4%	31% Acanthocytes	
MCV	84.5 ft	(wet smear examination) (control - 3%)	
MCH	28.3 Pg	normocytic, normochromic blood picture	
MCHC	33.5%	Blood group B+ve	
ESR	02 mm/hr	Reticulocytes < 1%	
WBC-TC	8400/ul	Platelets 3.93 L	
		lipoproteins %	
Cholestrol	233 mg/dl	Alpha	22 (23+4)
Triglycerides	157 mg/dl	Prebeta	6.1 (12+4)
LDL	153 mg/dl pre	Beta	66.2 (65+8)
HDL	48 mg/dl	chylomicron 5.7	
		No abetaliproproteinemia	
ENMG	Findings suggestive of demyelinating senson - motor neuropathy		

and agnoists were not effective in preventing tongue injuries and tics. We tried lithium as it has been reported in literature that a combination of lithium and tetrabenazine is useful in the treatment of orofacial dyskinesia (Jankovic and Janet Orman,1988, Reches et al.,1983). The anxiety, depressive symptoms and chronic insomnia also improved with lithium. Lithium has not been tried in the earlier case reports of neuroacanthocytosis from India (Bharucha and Bharucha,1989, Murthy et al.,1994; Roy et al.,1999).

This case report highlights the significance of peripheral blood smear examination in all cases of severe tic and dyskinetic disorders.

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