IDIOPATHIC TORSION DYSTONIA WITH SCHIZOPHRENIA IN FIRST DEGREE RELATIVES: A CASE REPORT

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SUMMARY

A patient having two schizophrenic brothers developed simple writer's cramp at the age of 20 years. Three years later she developed irregular and unusual movements which was diagnosed and treated as hysteria until she had contractures in the right hand. EMG studies revealed abnormalities suggestive of torsion dystonia. The association of schizophrenia in the first degree relatives and the differential responses to haloperidol in this case of torsion dystonia has been discussed.

Idiopathic torsion dystonia, whether focal, segmental or generalized, has been often misdiagnosed as conversion hysteria in the past (Lesser and Fahn, 1978; Cooper et al., 1976). These abnormal movements and dystonic postures are still being treated as "Functional Psychiatric Disturbances" (Roth, 1980). In the context of focal dystonias, it should be noted that an occasional case of simple writer's cramp may progress to focal or generalised torsion dystonias (Marsden, 1976; Roth, 1980). The authors are reporting a case bridging these two syndromes, i.e., simple and dystonic writer's cramp—a rare transition hitherto not reported from India. The authors are also not aware of any case of idiopathic torsion dystonia with schizophrenia in other sibs, an unusual association in this case that stimulates a few hypotheses regarding its aetio-pathogenesis.

CASE REPORT

A 23½ year old lady, clerk in the postal department was admitted with the complaints of inability to write for the past 3½ years and involuntary movements involving the upper limbs for 6 months. Born of a consanguineous parentage, the patient had a normal birth and developmental history. The two elder brothers were suffering from chronic schizophrenia. There was no family history of movement disorder or dementia.

Her symptoms commenced insidiously 3½ years ago with difficulty to write and having to strain excessively to grasp the pen. Handwriting deteriorated gradually and she had to pause several times due to pain and stiffness of the hand and forearm. All other finer acts were preserved. Physical examination and investigations failed to implicate any organic aetiology. She was diagnosed as a case of writer's cramp (occupational neurosis). However, behaviour therapy and later, psychotherapy were not effective excepting initial, marginal improvement.

She maintained same status until two years later when her symptoms worsened. On any attempt to write, she would frequently drop her pen due to sustained flexion of the thumb occurring intermittently. Neurological examination was normal again.

Six months before admission (i.e., 3½ years after the onset of writer's cramp), she
developed additional symptoms of irregular involuntary movements of the upper limbs and stiffness of the right forearm; she had to be bathed, dressed and fed by others. On examination, she appeared to be highly attention-seeking. There were unusual, non-purposive, occasionally repetitive movements affecting both the upper limbs (Right more than left) and neck, which aggravated on suggestion, attention and sympathy and diminished much on diverting her attention. There was no neurological deficit. Investigations including copper studies did not indicate any organic basis.

After the initial evaluation in terms of history, mental state, neurological examination and investigations, a provisional diagnosis of hysteria was made. Attempts to explore the psychodynamics, including pentothal interviews were fruitless. Her movements aggravated on the days of grand rounds; in between the weekly rounds, her movements remained much less and sometimes totally disappeared while relaxing alone in bed. Psychotherapy and placebo preparations failed to improve her conditions. On the other hand contractures and abnormal postures appeared and prompted an EMG screening. This showed simultaneous contraction of both the agonist and antagonist muscle of the right forearm. Hence her diagnosis had to be revised in the light of this evidence, suggesting an organic basis. She was diagnosed as a case of “late-onset” idiopathic torsion dystonia according to Marsden et al.'s criteria (1976) having grade 5 disability (Bundey et al., 1975).

Although adequate trials with L-DOPA and later, with carbamazepine were unsatisfactory, haloperidol seemed to be effective in comparatively higher doses. In a double-blind cross-over trial with I.V. haloperidol Vs. 25% Dextrose, there was significant improvement with haloperidol both in frequency and severity of the movements. Hence, she was started on oral haloperidol 1.5 mg/day, stepping up to 15 mg/day. But for the contractures and flexion deformities, the movements were controlled satisfactorily with the high doses which she tolerated well. With physiotherapy, flexion deformities responded slowly over a period of one month. As she could attend to all her daily activities independently including writing very slowly, she was discharged with grade 2 disability (Bundey et al., 1975). She maintained improvement at subsequent follow ups.

DISCUSSION

In this patient, initial involvement of only the singular act of writing, bizarre morphology of the movements, demonstrative behaviour, absence of any neurological deficit, her sensitivity to mental and social stresses and disappearance of the movements often when alone in the ward—all these have understandably misled the team to consider hysteria as the provisional diagnosis. In this context, the present case also highlights the importance of electromyographic studies which can detect simultaneous contraction of agonists and antagonists (Von Reis, 1954) in these cases and prevent “psychofication” of a treatable organic illness (Heilbrunn, 1978).

However, schizophrenia in two brothers and her differential response to higher than usual doses of haloperidol has been thought provoking. Also, she tolerated this high dose with least extrapyramidal and other side effects. The authors wonder whether torsion dystonia shares similar biochemical lesion with schizophrenia, where dopaminergic systems are implicated. It is tempting to hypothesize differential involvement of the same lesion giving rise to these two syndromes—a possible mesolimbic dysfunction in schizophrenia (two brothers) and nigro-striatal idiosyncracy in torsion dystonia (the patient). So far, results of CSF biogenic amine studies in torsion dystonia have been inconclusive (Allen and Knopp,
1976) yet, there is scope for further studies, especially linking the biochemical basis of torsion dystonia with that of schizophrenia. However, caution should be exercised in hypothesizing from a single case material, more so because of inability to do CSF studies in this patient due to lack of informed consent. Hence, the authors are currently investigating polled writer’s cramp cases with regard to their clinical profile, course, EMG pattern, dopamine-beta-hydroxylase activity and other variables in order to understand this clouded area at the interface between psychiatry and neurology and therapeutic implications of various sub-syndromes grouped under torsion dystonia.

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Appendix 1

Bundy et al.’s Criteria for Assessment of Functional Disability :
Grade 1 : Leading normal life ; symptoms.
Grade 2 : Mild disability ; continuing full-time work.
Grade 3 : Moderate disability ; works with difficulty.
Grade 4 : Severely disabled ; not at work ; independent at home.
Grade 5 : Wholly independent upon others.

Appendix 2

(a) Marsden et al.’s Criteria for Idiopathic Torsion Dystonia :
(1) The presence of dystonic movements and postures (but arbitrarily excluding isolated spasmodic torticollis).
(2) Normal perinatal history and early development.
(3) No history of any known precipitating illness or exposure to drugs known to provoke torsion dystonia prior to the onset of the disease.
(4) No evidence of intellectual, pyramidal, cerebellar or sensory deficit on clinical examination.
(5) Failure of laboratory investigations, including copper studies to demonstrate any cause for the disease.