Torsion Dystonia:
A Case Report

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A 35-year-old black female with typical torsion dystonia is discussed. Tremors in the right upper extremity began with a febrile illness at age eight. Difficulty in using the extremity began two years later. Overt writhing movements and torticollis began at age 17. The disorder has been progressive, but not disabling. Neurological examination revealed only extrapyramidal motor system dysfunction. Serum dopamine Beta hydroxylase levels were normal, and an evaluation for Wilson's disease was negative. A sibling has minor writing difficulties.

Torsion dystonias occur in two major patterns: the specific, often familial entity "dystonia musculorum deformans," and as "symptomatic dystonias," which occur secondary to identifiable neurologic disease.

Dystonia musculorum deformans is an uncommon disorder which has been described[1-3] in perhaps 10 or 11 cases in people of African origin. The symptomatic dystonias have also been infrequently[2] reported (six or seven cases) in black people (Table 1).

The patient discussed here is a 35-year-old black female with torsion dystonia who displays some historical aspects of both the congenital and symptomatic disorder. It is proposed that the case represents an instance of acquired dystonia in someone genetically predisposed, in an autosomal-recessive pattern, to torsion dystonia.

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Case Report

The patient presented to the Neurology Clinic in early 1975 with uncontrolled movements of the right upper extremity and torticollis to the right.

She recalled having had a fever, headache, and sore throat at age eight and during that illness, slight tremors of the right upper extremity were noted.

During the episode she "saw things," though she was not drowsy and spoke rationally. She was seen in the hospital as an outpatient.

Subsequently she "saw things" intermittently with febrile episodes until age 16. The visual phenomena were of familiar people doing unthreatening and silly things.

From age eight to ten years, the tremor in the right upper extremity was present. At age ten, she first noticed trouble in writing with the right hand; this was sufficiently intense that she switched to using the left hand. Writhing movements and neck torsion to the right began at about age 17. The neck torsion has been steadily progressive.

About five years ago, the motor problems worsened, concomitant with domestic problems. The worsening has persisted.

Birth had been normal, as were growth and development before age eight. Sarcoidosis was diagnosed in 1968, but she was not treated.

The patient had previously been observed in two university hospitals because of her motor problems. During a 1973 admission, she had a pneumoencephalogram and a brain scan; both were reported as normal.

The patient has no children. She is the oldest of five girls and one boy. A 30-year-old sister has some writing troubles. "When she writes it looks trembly." This has been present intermittently (most of the time) since about age 14 and is not worsening. She must write more slowly when having the problem, although the writing improves somewhat after she writes for a while.

The father had spinal meningitis 30 years ago. He was unconscious for 14 days and has since walked with a limp.

The patient functions adequately in a full time clerical post.

Table 1. Torsion Dystonia in Blacks – Total 19 Cases

Reference	Relevant Illness	Familial Factor	Other
Austregesilo,[9] 1934	Sleeping sickness	–	2-year-old child
Elfenbein,[10] 1968	Lipid disease	Yes	Disease in 3 relatives
Weir, 1976	Febrile illness with visual phenomena	Yes, 2 sisters	(Torsion dystonia) Writing difficulty
Zeman and Whitlock,[11] 1968	Urinary tract infection at age 3	–	20-year-old Jamaican (racial identity not certain)
B.J. Alpers (Ciba film)	–	–	Dystonia musculorum deformans (unreported case)
Phelps,[13] 1961	–	–	Male (symptoms in leg trunk)
Zeman and Dyken,[5] 1968	–	Yes	Father and 2 sons
*Golden,[2] 1976	–	Mother with psychiatric illness	Patient is mentally retarded
3 sporadic cases (in survey by Golden)	–	–	Onset in first decade
2 familial cases (in survey by Golden)	–	Yes	Patient and grandfather
Dr. Miller (discussion of paper by Dr. Golden)	–	–	–

*In a recent review on dystonia in blacks and Puerto Ricans.

Examination*

There was no abnormality in mental status, cranial nerves (note torticollis below), cerebellar system, sensory system, or the reflexes.

The head was rotated to the right most of the time, although it often spontaneously returned *almost* to the midline. A moderate scoliosis with torsion inferiorly to the right was present, and the right shoulder girdle protruded posteriorly. Marked writhing movements of the right upper limb, and less often in the left upper limb, were present. Only occasional movements of the right lower limb were noted. The movements were somewhat greater proximally than distally.

Motor tone appeared normal except for an increase at the right wrist. Strength was intact and gait seemed impaired only by truncal torsion. No tremor was detectable clinically, although one was subsequently recorded on EKG.

A sample of spiral drawing was obtained in early 1975, and several further samples have been recorded (Figure 1).

Family Examination

The patient's mother and brother had normal neurologic examinations. The father (meningitis history) displayed a mild postural tremor of the hands, slight tremor of the tongue, and impaired bilateral foot tapping. A writing sample was obtained from the

*A five minute film is available from Dr. Roger Weir, Department of Neurology, Howard University Hospital, 2401 Georgia Avenue, NW, Washington, DC 20060.

affected sister (Figure 2). It was moderately different from an average writing sample but the rest of her neurologic evaluation was unremarkable.

Laboratory Data

Normal values included: serum dopamine β hydroxylase (DBH) of patient, mother, and sister; urine copper; serum ceruloplasmin; serum uric acid; SMA_{12}; Na; K; Cl; CO_2; urinalysis; and complete blood count. The serum copper was also normal. Chest x-ray showed sarcoid tubercules. Slit-lamp examination revealed no Kayser-Fleischer ring. Several indicated tests could not be performed because of movement interference.

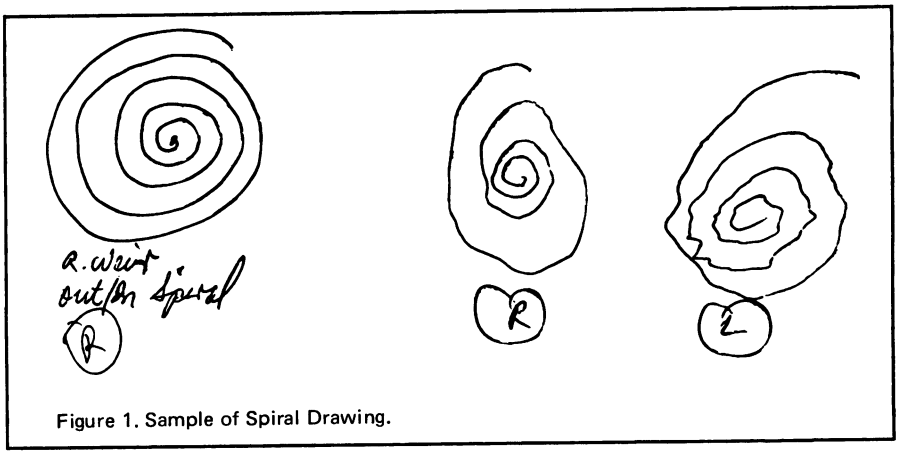

Figure 1. Sample of Spiral Drawing.

Discussion

The febrile illness at age eight appears to have been the prime etiology of this disorder. However, the mother indicated that the trouble *using* the right hand was the initial sign of illness; she only recalled the febrile episode on prompting.

Torsion dystonia,[4] secondary to infection but with no accompanying dementia, has been described. Similary, progressive dystonia[5] secondary to brain damage has often been recognized.

The family history of something akin to writer's cramp (a forme fruste[5,6] of dystonia musculorum deformans) and also of tremor and impaired foot tapping (although of unclear etiology) suggests the possibility of a hereditary dystonia. Elevated serum dopamine-β-hydroxylase levels would be consistent with dystonia musculorum deformans occurring in an autosomal-dominant[7,8] or an autosomal-recessive[8] pattern. The normal levels have no specific diagnostic value. The subsequent medical course may clarify etiologic factors in this case.

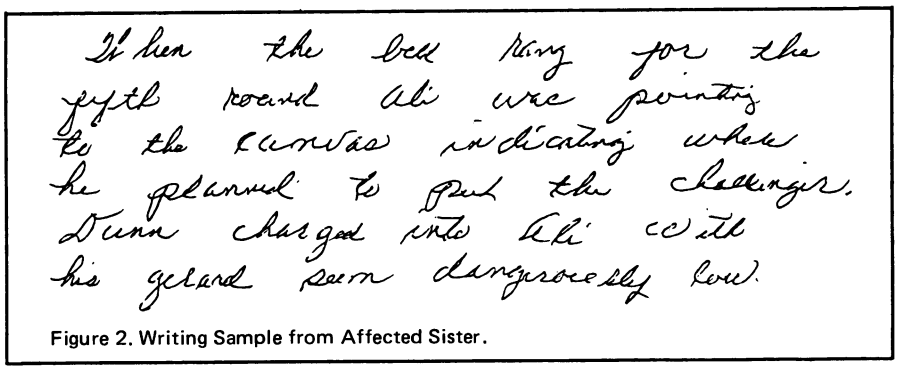
Figure 2. Writing Sample from Affected Sister.

Addendum

The patient has shown a positive therapeutic response to combination therapy with diazepam (Valium), clonazepam (Clonopin), and biperiden (Akineton). It appears that at least Valium and Clonopin are necessary for this response.

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