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Hypomanic Behaviour Associated with Familial Spastic Paraplegia

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Summary. Psychiatric manifestations of familial spastic paraplegia are rare and have been described only infrequently. A 35-year-old male is reported, who presented both hypomanic behaviour and gait disturbances as features of a previously undiagnosed familial spastic paraplegia. This association implies that the CNS manifestations of familial spastic paraplegia may overlap with the neurochemical or neuroanatomic substrata regulating mood.

Key words: Familial spastic paraplegia – Hypomania

Introduction

Familial spastic paraplegia (FSP), as first described by Seeligmuller (1876) [10] and Strümpell (1880), [11] is primarily a progressive spinal cord disorder with pyramidal tract involvement, resulting in spastic gait. Inheritance is usually autosomal dominant although recessive forms occur; sex-linked inheritance and sporadic cases have been reported [13]. The expression of the disease varies and even members of the same family may show a wide variety of signs and symptoms.

Although the disease may exist in a pure form, showing pyramidal signs only, other signs and symptoms such as ataxia, dementia, extra-pyramidal signs, optic atrophy, muscle wasting and posterior column involvement have been described along with skeletal and cardiac abnormalities [4, 9, 13]. These variants are of importance in order to understand the extent of this comprehensive disorder and in establishing it as a nosologic entity. The age of onset of FSP differs considerably and ranges from infancy to senescence. The age of disease manifestation may be constant within one sibship; this, however, has not been confirmed by some authors [4, 8]. The recessive form of the disease usually commences at an earlier age and progresses more rapidly than the dominant form [4, 8, 13]. A sex predilection of slight degree for males was found in both autosomal dominant and recessive FSP [1, 8].

Diagnosis of FSP is based on the concurrence of clinical signs and symptoms upon physical examination and a positive family history. Diagnostic criteria at physical examination are not universally accepted. The presence of exaggerated tendon reflexes only is considered evidence of incipient disease by some, but not by others.

Bickerstaff requires the exaggeration of tendon reflexes to be very marked and accompanied by some other feature or features of the fully established condition, e.g. the characteristic foot deformity or abnormal plantar responses [1]. Considerable overlap of symptoms exists with other hereditary degenerative neurological diseases, e.g., spinocerebellar degeneration, Friedreich's ataxia, familial forms of amyotrophic lateral sclerosis. The ultimate classification of these disorders awaits elucidation of the basic underlying biochemical abnormalities. We describe a patient who presented with serious psychiatric symptoms and subsequently appeared to be the proband of a three-generation kindred with FSP.

Case Report

Mr. A, a 35-year-old white male teachter (III–3 Fig. 1) had been suffering from a progressively stiff gait and an dysinhibited urinary bladder for a period of 4 years. For the previous 2 years his family had noted a severe personality change from a stable mood disposition into euphoria, irritability with sudden emotional outbursts and a tendency to hypersexuality. Mr. A denied a personality change but admitted that his behaviour interfered with his professional activities and caused marital conflicts. Pupils accused him of dipsomania because of his gait

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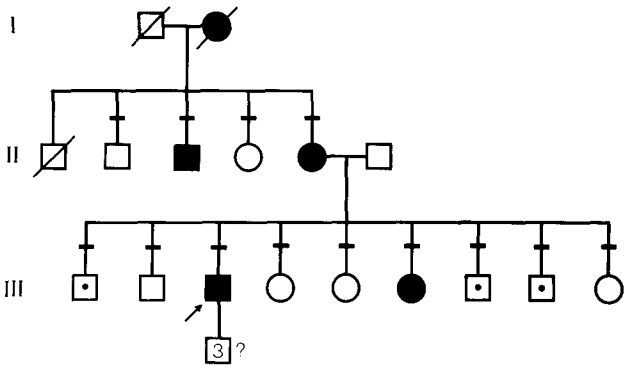

Fig. 1. Three-generation family tree presenting FSP (*shading* = clinically affected patient; *horizontal bar* = personally examined; *central dot* = minimally affected; *circles* = females; *squares* = males; *number in symbols* = number of offspring; *arrow* = proband; *oblique bar* = deceased)

disturbances, high spirits and loud voice. Friends and acquaintances avoided his irritant, often provocative manners. His history revealed no bipolar episodes or other psychiatric manifestations, and for the previous 10 years he had worked steadily at the same job. Mr. A took no medication or drugs and never had more than two alcoholic drinks a day.

On physical examination Mr. A showed a characteristic scissors gait with marked spasticity of the legs and relatively little loss of muscle power. Knee and ankle jerks were pathologically brisk and plantar responses were extensor on both sides. There was some impairment of vibration sense in the lower limbs with an impaired position sense of the toes.

Pes cavus was bilateral with the arching disappearing on standing. Laboratory investigations of CSF, blood and urine were normal; lues serology was negative; electroencephalography, electromyography, myelography, cerebral nuclear magnetic resonance scanning and cerebral computerized tomography with and without enhancement were negative.

During the psychiatric examination Mr. A was restless and cheerful and walked away repeatedly. He was inappropriately unconcerned and spoke almost continuously in a loud voice, abruptly changing from one subject to another. Flight of ideas was prominent and witzelsucht was conspicuous. His attention was easily distracted by extraneous stimuli. His mood was uniformly euphoric, except for occasional outbursts of temper. There were no changes in alertness, signs of intellectual deterioration or memory loss. Criticism by his wife and other persons was minimized, often in a tactless and inconsiderate way. There were no delusions, hallucinations or disorientation.

Mr. A was the third of nine children, the offspring of nonconsanguinous parents of Dutch ancestry. A sister of Mr. A (III–6 Fig. 1) is known to suffer from periodic euphoria and hyperactivity.

Family investigation produced a three-generation family tree, clearly exhibiting a previously undiagnosed FSP with reduced penetrance (Fig. 1). Onset of FSP in this family varied between adolescence and early adulthood (18–22 years).

The three sons of Mr. A are too young to assess the possibility of their being afflicted by FSP. From the pedigree described, inheritance was not necessarily autosomal dominant. No example of male to male transmission is known to occur, so that some form of X-linked inheritance is possible.

Table 1. Various clinical manifestations of familial spastic paraplegia (FSP) (after Sutherland)

Pure FSP (Strümpell's disease)	
FSP with associated features:	– impaired intelligence
	– optic atrophy
	– impaired vibration sense
	– ataxia
	– pes cavus
Complicated FSP:	– hypomania
	– dementia
	– extrapyramidal features
	– retinal degeneration
	– amyotrophy
	– cardiomyopathy

Discussion

This study highlights diagnostic difficulties encountered in FSP. The disease may be disguised because of its protean manifestations and varying degrees of severity, even in members of the same family. Sutherland suggested referring to pure FSP or Strümpell's disease; FSP with associated features; associated features only without FSP; complicated FSP with associated features unusually well-marked or encountered only rarely and finally of complicated FSP with associated features only [13] (Table 1).

Even in outspoken, fully developed cases of pure FSP the diagnosis might be missed because of the patient's acceptance of his very slowly progressive stiff gait, blaming it on old age and not seeking any medical help. Associated features as the only manifestation of the disease without spastic paraplegia may easily be misinterpreted if a positive family history is lacking. Unawareness by the patient that the disease may be familial and hereditary, especially in the autosomal recessive and sporadic forms, is an additional factor that interferes with a positive family history.

At physical examination of patients suffering from FSP the so-called classical pes cavus often is not typical, as the antero-posterior shortening of the foot and expected concavity of the plantar surface is reduced and apparently filled in by soft tissue when the patient is standing.

The discrepancy between an often severe spasticity of the legs and well-preserved muscle power with only slight paraparesis may form another clinical pitfall. A real paraplegia may be lacking in autosomal dominant FSP, whereas the recessive variety may run a more severe course. In some patients spasticity of the legs is such that tendon reflexes cannot be elicited and are incorrectly documented as being absent.

Superficial abdominal reflexes are often preserved and bladder and bowel function is mostly normal. Even extensor responses may be absent for a relatively long time as in FSP the medial vestibulospinal and reticulospinal tracts may be more affected than the lateral corticospinal tracts [2].

Serious pitfalls in the diagnosis of FSP are sometimes caused by the association of features that are observed only rarely in this disease. The association of psychiatric symptoms with FSP is rare. Bickerstaff reported euphoria as an outstanding sign in several affected individuals of a family with autosomal dominant FSP [1]. Sutherland described frank euphoria, dulling of intellect with obsessional trends and presenile dementia in a family with autosomal dominant FSP [12]. In the Ferguson-Critchley syndrome, consisting of hereditary spastic paraplegia with ocular and extra-pyramidal signs, euphoria was described repeatedly [3, 5, 7].

The psychiatric condition of Mr. A met the DSM III criteria for an organic personality syndrome with a predominantly hypomanic mood disturbance (organic affective syndrome). Manic psychosis and frontal lobe syndrome were initially considered as possible causes of the patient's outstanding hypomania. Only the finding of spastic paraparesis upon neurological examination and the positive family examination indicated that his behaviour was not purely coincidental but an associated feature of FSP. This finding might influence therapeutic guidelines as the mental abnormality is organically determined. No specific organic substratum for hypomanic or manic mood disturbances is known, although limbic pathology is sometimes referred to [6]. The association between FSP and hypomania, as presented in this case report, implies that the CNS manifestations of FSP may overlap with the neurochemical or neuroanatomic substrata regulating mood.

However, neuroradiological, neurophysiological, neuropsychological and neurochemical assessments did not elucidate the exact mode in which the two symptom complexes are linked. The currently understood body of knowledge regarding either disorder still awaits further clarification of basic underlying biochemical abnormalities.

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