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Huntington's Chorea without Dementia
A Problem Case

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Summary: An unusual case of Huntington's Chorea, without dementia, is described. The diagnostic issues are discussed with particular reference to the problems of psychiatric assessment and management of the disease.

Huntington's Chorea is generally regarded as the classical autosomal inherited illness. Routine teaching holds that the gene is dominant, penetrance is full, the movement disorder typical, and the mental decline invariable. Psychiatric symptoms are very common and undiagnosed cases usually found in mental hospital wards. However "the rich gamut of clinical manifestations makes the apparently simple diagnosis exceedingly difficult in some instances" (Bruyn, 1968), and the absence of dementia though described, without references, in several major textbooks, (Brain & Walton, 1969; Lishman, 1978), seems to be rare indeed. Despite several false hopes, there is no accepted diagnostic test of the condition apart from post-mortem analysis of the brain, while treatment of the condition is very limited. Although it seems clear that a disorder of GABA-ergic and cholinergic neurons is involved, (Marsden, 1982), the ethics of a predictive test, (Thomas, 1982), and the role of genetic counselling, (Harper *et al*, 1981), have assumed a primary importance, for the present, in the practical management of the illness. The case reported here illustrates these issues very well; the patient is not dementing, the family history is uncertain, and the relatives are divided in their attitudes.

Case history

The patient, a 54 year old married man, a builder and carpenter by trade, was referred for assessment (to the Maudsley Hospital) because of difficulties in diagnosis and management. He had a nine year history of recurrent manic-depressive illness, necessitating repeated admissions to hospital, sometimes under section orders. He sustained a fractured skull early in his illness (due to an accident on the railway that may have been a suicidal attempt) but recovered fully without neurological impairment. Some two years after this he was noted to have "semi-purposive movements" of his limbs and facial muscles, and these have persisted ever since, despite a range of treatments. These included drugs such as bromocriptine and levodopa, sodium valproate and brief courses of oral Haloperidol. Neuroleptics had not been used prior to his movement disorder, nor had he required high doses or long-acting preparations of such drugs at any time during his illness. His mood swings were eventually well controlled on lithium carbonate, but discharge home and a return to regular work were prevented because of his persistent argumentativeness, and social embarrassment occasioned by mildly aggressive outbursts. His full scale IQ never changed from approximately 120 on the WAIS, which was consistent with his premorbid abilities. His siblings were adamant that he was as bright as he had ever been. They could not understand why his wife would not have him home on a regular basis, and described *her* as "highly strung".

Full assessment on admission confirmed the physical and mental state findings. He had persistent choreiform movements, regarded by two experienced and senior neurologists as "typical of Huntington's Chorea". Routine tests of blood and urine were normal, including serum caeruloplasmin, copper and thyroid function. A full scale IQ of 121 was obtained on the WAIS with no evidence of decline or of significant cognitive deficits. Both EEG and CAT Scan were reported as normal, although it was conceded by the radiologists that the latter *might* show evidence of mild frontal lobe changes. His family confirmed some personality change in the direction of mild disinhibition, but examination of his mental state showed no overt affective, cognitive or perceptual disorder.

Follow-up two years letter showed no change, although there was some improvement socially. He was living independently in a hostel and attending a day centre, spending weekends with his wife and family. There had been no further incidents of disturbed behaviour, and his mood swings were very well controlled on regular lithium.

Family history

Further investigation of this family history revealed that his father had died aged 45 in a mental hospital, and the death certificate gave "Huntington's Chorea" as the cause. Descriptions from old medical notes described him as having "exaggerated movements of legs and arms", and "as hysterical in gait and manner as ever he was". However, no details were available of a post-mortem examination, nor was there any clear evidence of the course of illness. Furthermore, the paternal grandfather had died in a mental hospital, of a dementing illness, after being an in-patient for thirteen years, but his records were unavailable. However, the father's three brothers had apparently lived till their seventh decade, without any hint of movement disorder or mental disorder, and the patient had seven living siblings, aged between 42 and 60, all of whom were well. Interestingly, two others had died of

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neurological illnesses; an infant boy, aged two, died of "meningitis", while a sister died of pneumonia, secondary to severe multiple sclerosis, when aged 43.

Management

Despite the atypical features, the balance of evidence seemed to favour the diagnosis of Huntington's Chorea. But even the remote possibility of such a diagnosis met with considerable resistance from the extended family. A meeting was arranged to discuss the situation in full with the patient's wife and children, and three of his siblings. The uncertainty surrounding the diagnosis, and the possibility that this might be Huntington's Chorea in a most atypical form, were explained in some detail. Possible genetic risks to his children were described. His siblings were reassured that in all probability they had outlived the major risk period and were unlikely to have passed on the disease to their own off-spring. This explanation was accepted in general, and a degree of reconciliation reached. However, only the patient's own daughter made a firm decision not to have children. Subsequently she broke off her engagement. No other decisions of this nature have been reported since then.

Discussion

While the chorea, the father's diagnosis and the social and behavioural decline were all typical, the absence of dementia and the presence of nine unaffected siblings were puzzling. The significance of the head injury was discounted. This factor had long been blamed by the patient's siblings for his subsequent decline, while gas poisoning in the First World War was the family's explanation for the patient's father's illness. However, the length of time between insult and the development of chorea, in both instances, was strongly against such an interpretation. As to the unaffected siblings, Mendelian genetics predicts a 50% incidence of chorea; yet three out of four brothers in the father's generation remained well, as did nine out of 10 siblings in the patient's generation. Possibly the two siblings who died (aged two and 42) had atypical forms of the illness (Oliver and Dewhurst, 1969); also several of the younger ones (in their 40's) may yet develop symptoms. Age of onset in the fourth and fifth decade is typical but not absolute.

The absence of dementia is extremely rare, the few reports available all describing an onset in early childhood, (Behan and Bore, 1977). In one large series (Heathfield, 1967) there is a description of a 60 year old woman with severe chorea for nine years, who was still living at home and without apparent dementia. It may yet develop in the present patient, although a gap of 10 years would be extremely unusual. He has certainly had clear-cut psychiatric symptoms and a degree of change in his behaviour and personality, but these do not seem sufficient to accord with the classical diagnosis. Although typical cognitive impairment is described, (Aminoff *et al,* 1975), a whole range of psychological disturbances have been reported in larger series (Brothers & Meadows, 1953). It might be argued that the word 'dementia' has been used in the older literature to describe a combination of personality changes, psychiatric symptoms, and even the effects of serious drug or alcohol abuse. However, detailed studies seem to have excluded such a possibility. (Heathfield, 1967; Bolt, 1970).

The essence of the dilemma in this case was how to approach the relatives. The patient had three children in their teens; the eldest had just become engaged to be married. There are altogether 19 nephews and nieces, four from the sibling with multiple sclerosis. Given our doubts about the diagnosis, how fair was it to advise having children? Genetic counselling does seem effective, (Harper *et al,* 1981), but many would question the ethical side of such management (Thomas, 1982) even when diagnosis is certain. Our solution was to discuss things openly in a family meeting, explaining areas of doubt and uncertainty, with the outcome as described above.

Apart from the poverty of our diagnostic assessments, two matters of importance seem to emerge from this case. First, there is a need for more frequent post-mortem analyses of the brains of psychiatric patients having distinct organic features. Secondly, a more detailed analysis of the incidence of non-dementing chorea may prove fruitful. Distressing though the choreiform movements are, it is the dementia that provokes much of the personal and family misery. Its absence or delay in onset may be an indication for alternative policies in management and counselling.

Finally, some might suggest that this patient does not have a true Huntington's Chorea. There are several inherited movement disorders that can mimic the condition. The patient's head injury may have been significant. We were however assiduous in obtaining experienced neurological assessment, and alternative causes of chorea (e.g. Wilson's disease, cerebral lesions) were carefully excluded. Given his history, a drug-induced dyskinesia was also discounted. In the end, the diagnosis can only be made clinically, and all clinical opinions concurred that this was Huntington's Chorea. It is in our understanding of the illness that the problems seem to lie, and these are highlighted by this case.

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A 'Pseudo-AIDS' Syndrome following from Fear of AIDS

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We report two cases showing psychiatric symptoms associated with a fear of Acquired Immune Deficiency Syndrome (AIDS). There have now been over 42 cases of AIDS in the United Kingdom (Communicable Disease Surveillance Centre, 1984) since the first case appeared in December 1981, a year after the syndrome was first identified in the United States of America. It was the BBC television programme "Death in the Village" which brought the risks home to many UK homosexual men in mid-1983. Following the programme, the London Gay Switchboard received a high level of telephone calls from men who were afraid that they had AIDS (London Gay Switchboard, 1984).

The early symptoms of AIDS are very similar to those of anxiety and depression. The AIDS sufferer feels lethargic, loses appetite and weight, and sweats excessively (Foege, 1984). Diagnostic complications arise from there being no unique diagnostic test for the condition. Immunological tests, in combination with other evidence, are only suggestive of the presence of AIDS, and the diagnosis can only be confirmed by the finding of Kaposi's sarcoma, Pneumocystis carinii pneumonia, or other persistent opportunistic infection. Therefore, it is not possible to be certain that a patient has AIDS in the early stages, nor is it usually possible to give someone an absolute assurance that he does not have it, until the definitive diagnostic criteria have been eliminated. This in itself can be a problem, as the incubation period is over twelve months in duration. When combined with the degree of misinformation that is often present in lay understanding of public health issues, there may be considerable misinterpretation and anxiety when innocuous or coincidental signs appear.

Case 1: Depression

The patient, a 25 year-old homosexual male, was referred to the psychiatric department from the venereology clinic. He had presented there with the belief that he had contracted AIDS as the result of a casual contact with an American male, six months previously. He remained convinced of this after extensive negative investigation, including skin biopsy, and reassurance from a senior consultant venereologist. There was a three-month history of depressed mood with diurnal variation, early morning waking, weight loss of 5 kg, loss of interest in his work, and malaise. Additionally, the patient had barely discernible capillary haemangiomata on his arms, upon which he carried out ritualistic self-examination, on average six times daily. All of the symptoms were interpreted by the patient as being indicative of AIDS. He spent several hours praying each day, considered his situation hopeless, and recently made a will, and planned suicide by self-poisoning. Since the age of 16, he had had

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