Huntington's Chorea in a Chinese Family

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Since 1872 when Huntington first described the condition which has borne his name the disorder has been reported in a number of races. The first Chinese family, in which eight cases were affected through four generations, was reported by Singer (1962) from Hong Kong. The present paper represents the first report of such a family from Formosa. In all, 11 persons in 4 generations are known to have been affected, as may be seen from the diagrammatic pedigree (Fig.) and the descriptive appendix; clinical findings in 4 who were still alive at the time of the examination are described below.

Case Reports

Propositus (III.4). A 40-year-old man was admitted to the Department of Neurology and Psychiatry, National Taiwan University Hospital, Taipei, in February 1966. He had been born in a small farming village in northern Formosa, and had remained well until the age of 35 years when he noticed that his right small finger occasionally twitched. At about the same time he began to be troubled by involuntary grimacing and shrugging of the shoulders. Within a year thereafter movements appeared in other fingers, arms, and legs, and he had slight twisting of the trunk and neck when he walked. At 37 the twitching became constant and his speech unclear so that he had to resign his teaching post at a primary school. Then he managed a small grocery shop fairly well for nearly a year, but began to complain of difficulty in concentration and of forgetfulness. About a year later he became careless of his personal appearance and the twitching became more severe.

On admission he looked dirty and slovenly. There was frequent writhing contortion of the facial muscles, involuntary jerky movements of the hands, limbs, and shoulders, and flexion or extension of the trunk. These signs were increased in intensity by voluntary movements. The gait was ataxic, with alternate forward and backward lurching. His speech was dysarthric, with an abrupt staccato quality and rapid modulations in tone and tempo. He was generally apathetic and had difficulty in abstract thinking, but he did not have delusions or hallucinations.

Intellectual function and visuo-motor co-ordination were impaired. The short form WAIS verbal IQ was

56 (performance part impossible owing to twitchings). The span for immediate recall was very narrow; verbal abstract thinking was very poor, with distracted attention, and numerical performance was fair but poor relative to his educational background. The Rorschach Intelligence Index was (-9), which was very low for his past intellectual achievement; his thinking was retarded and stereotyped and his memory greatly impaired. The

FIG. Family pedigree

- I.1: Onset at about 40, died at 50; some of her sibs said to be affected?
- II.1: Died at about 60, not affected; his descendants not affected.
- II.2: Onset at 45, symptoms said to be more severe, died at 57.
- II.3: Onset at middle age; no children; died in his fifties.
- II.4: Died at about 60, not affected; two children: one male, not affected; one female, died in early childhood.
- II.5: Age 70, healthy; her descendants not affected.
- II.6: Onset, after marriage; expelled from her husband's home; had a child with a beggar but the child died in infancy; age at death not known.
- III.1: Onset at 28, died at 45.
- III.2: Onset at 27, died at 40.
- III.3: Died in infancy.
- III.4: Onset at 35, death at 41 (see text).
- III.5: Died in infancy.
- III.6: Age 37, healthy at present.
- IV.1: Age 24, onset 16 (see text).
- IV.2: Age 21, not affected at present.
- IV.3: Onset of minor jerky movements and stiffness of extremities at 14 followed by progressive deterioration of mental function, died at 22.
- IV.4: Age 24, onset at 18 (see text).
- IV.5: Onset at 16, death at 19 (see text).
- IV.6: Age 16, healthy at present.

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Bender-Gestalt test showed figure rotation, perseveration in figure copying, and poor visuo-motor co-ordination.

Routine blood chemistry and CSF examination showed no significant findings. ESR was raised (1 hour, 40 mm.; 2 hours, 81 mm.). ECG showed conspicuous left axis deviation and incomplete right bundle-branch block. EEG at rest showed a diffuse low voltage flat pattern with total absence of alpha rhythm, and the only waves seen were of low voltage, 20 microvolts, and 24–28 c/s rhythm. Pneumoencephalogram showed dilatation of both lateral ventricles without shifting, and there was irregular filling of the sulci at the frontal, parietal, and temporal regions, as well as basally.

Chlordiazepoxide 30 mg. and thioridazine 75 mg. daily were given, and the choreic movements moderately improved about one week after the medication. At discharge, one month after admission, he was able to sit quietly for a while, and his apathy somewhat improved, but the dementia remained unchanged. He died in April 1966 from accidental head injury at the age of 41. The family refused necropsy.

IV.1. This male patient, born in 1941, had been physically weak, passive, introverted, and quick tempered from childhood. He had made poor grades at school and had always felt inferior. After school, at the age of 14, he helped the family business dealing in ducks. At 16 he began to notice clumsiness of the right hand and some difficulty in memory, and became depressed and sleepless. At the age of 18 he began to have occasional jerky shaking of neck and shrugging of shoulders. He continued to be depressed, and had suicidal ideas, difficulty in concentration, and occasional severe insomnia. At 23 he attempted suicide with sleeping pills.

On examination at the age of 24 years he was depressed, had ideas of self-depreciation, hopelessness, and suicide; speech was slow, voice was low, with occasional stuttering. Movements were generally slow, with occasional sudden shaking of neck and shrugging of shoulder. He had no delusions or hallucinations.

His WAIS performance IQ was 70, and his verbal IQ 87. There was evidence of impairment of intellectual function, particularly in abstract thinking, perceptual organization, and motor speed. The Bender Recall Test indicated gross visuo-motor dysfunction.

He was treated with diazepam 15 mg. daily and was advised to report regularly but did not return.

IV.4. At 18 years this male patient, born in 1944, began to stutter occasionally and to walk awkwardly with propulsive gait. A year later he began to have occasional jerky movements of the limbs and difficulty in concentration and memory. Thereafter he deteriorated rapidly, though his physical condition remained nearly stationary.

Examined at the age of 24, he appeared very dull, with frequent blinking of eyes, marked rigidity of extremities, and occasional sudden nodding and jerky movements of the fingers, but he was still able to walk without support and to feed himself fairly well. He was able to respond

to questions by explosive utterance of simple sentences only with great effort. No gross disorientation to time, place, and person. In a 100–7 test he was unable to proceed beyond 93. Remote memory was fairly well preserved, but recent memory, ability to retain immediate visual and auditory stimuli, and abstract thinking were greatly impaired. Further investigation was refused.

IV.5. This man, born in 1947, first noticed occasional jerky movements of limbs and trunks at the age of 16 years. Subsequently his mental functions and speech deteriorated very rapidly. By the age of 18 he was no longer able to walk or feed himself because of stiffness of the extremities.

Examined at home a year later he was apathetic, dirty, and totally demented, and he could only utter a few explosive unclear words in response to questions. He had marked rigidity of the extremities and trunk and could walk only with support with an unsteady propulsive gait. Occasional slight twitching of his fingers was noticed. He died at the age of 19 years.

Comment and Discussion

The family history is characteristic of dominant autosomal inheritance, and the whole course of the illness and the clinical appearances in the *propositus* are typical of Huntington's chorea. In his case, as well as in the elder members of the family who are all dead, the onset was in middle life and there is nothing obviously atypical. However, all his four nephews fell ill very young, between 14 and 18, and three of them showed unusual pictures. In the case of IV.1 the profound and lasting depression is noteworthy; and in IV.4 and IV.5 the very severe rigidity predominated.

The rigid form of the disease was reported as early as 1908 by Hamilton, and has been estimated to constitute 12 to 14% of all cases of Huntington's chorea (Panse, 1942). Their mean age of onset, 22.5 years, is earlier, and the duration of the disease, 11.1 years, is lower than those of the general run of cases (Bittenbender and Quadfasel, 1962). In the present cases, IV.4's illness took place at 18, with steady deterioration manifested by rigidity and dementia when he was examined 6 years later at 24. IV.5 became ill very early at 16, and took a rapid deteriorating course, with marked rigidity and dementia, resulting in death at 19, 3 years after the onset. It is noteworthy that the early onset and the rigid form, as found in more than one case in the present family, are reported by Bittenbender and Quadfasel (1962) and Jervis (1963).

Summary

The present paper is the first report of Huntington's chorea in a Formosan Chinese family. There

were 11 cases known to be affected through four generations. Case histories of four affected patients who were still alive at the time of the examination have been described: though the propositus showed typical pictures of progressive chorea and dementia starting from middle age, the other three cases became ill very early, from 16 to 18, with main features of depression in one and marked rigidity and progressive dementia in the other two. It is noteworthy that the early onset of the illness and the rigid form were found in more than one case in the present family, as has been reported by others.

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