UNUSUAL CASE OF TUBEROUS SCLEROSIS

BY

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[WITH SPECIAL PLATE]

Tuberous sclerosis is a condition found in the brain as a feature of the syndrome epiloia. Crichtley and Earl (1932) described 29 cases of epiloia, and in a review of the literature pointed out the essential features of the disease. Classical cases are characterized by mental deficiency, recognizable in early life, adenoma sebaceum at puberty, and epileptic fits. A psychopathic taint is often a feature of the mental state, and motor restlessness is common. A familial incidence may also be discovered. Other signs recognizable during life may include thickening (shagreen patches) of the skin, flattened dermal fibromata on the back, pedunculated dermal tags, and such stigmata as simian hands, with incurving of the little finger, and misshapen ears.

At post-mortem examination the cerebral convolutions are found to contain numerous sclerotic areas which on microscopy appear to consist of an increase in neuroglial tissue with the addition of giant cells foreign to the central nervous system. Tumours of other organs are common, including cardiac myomata and bizarre renal neoplasms varying from sarcoma to hypernephroma.

Tredgold (1947) described the condition and discussed the features of the mental state. Clinical variants occur, and Stewart (1935) recorded the sudden death in epilepsy of an apparently normal young woman. Typical sclerotic areas were found in the brain, though there was no previous history of mental deficiency or fits, and no evidence of adenoma sebaceum or renal neoplasms.

The purpose of this paper is to describe another of the *formes frustes* of epiloia which occurred in a young soldier.

Case Report

The patient was called up for national service in August, 1949. He was found fit and drafted to a selection unit. His medical documents stated: "Pigmented rash on face. Marked refractive error in both eyes—corrected to 6/6. A history of migrainous headaches." There was no history of fits of any kind. He underwent the usual selection procedure at his unit and was given the intelligence rating "S.G. 3P." This placed him in the top 50% of the Army population and showed him to be above average intelligence. The personnel selection officer made the following comments at a routine interview: "He is a man of above average intelligence and ability and has been on survey work. He is of negative personality."

It should be noted, however, that the man's instructors described him as "slow to learn," while his father later stated that although he was an affectionate boy he was not as quick as his brother. There was no family history of mental disease or of fits.

Soon after his arrival at the selection unit he reported sick with headaches. The presence of adenoma sebaceum on the face was at this time remarked upon and he was kept under observation. Four weeks later his comrades reported, at about 8.30 p.m., that he had complained of drowsiness at about 4.30 p.m., had gone to bed, and could not be roused.

On admission to hospital he was semi-comatose and restless, with alternating pulse and Cheyne-Stokes respiration. The limbs were in the position of decerebrate rigidity; the arms being

flexed and legs extended. The tonic state of the limbs was described as striking. The pupils were at first equal and reacted to light, but as his condition deteriorated into deep coma they became fixed and dilated. The tendon reflexes were brisk but the abdominal reflexes were absent.

He died at 10.45 p.m., apparently from a brain-stem lesion, about seven hours after he had first complained of drowsiness.

Post-mortem Examination

External Appearance.—The body was that of a well-nourished young adult male. The pupils were dilated and equal. There was a small pedunculated papilloma close to the left nasal cleft surrounded by several sessile red and brown plaques varying from a pin's head to 2 mm. in size. Similar plaques and a tiny papilloma were visible in the right nasal cleft. A few tiny red spots were visible on the cheeks. The distribution of these plaques was of a butterfly nature and the condition appeared to be one of adenoma sebaceum. There was a small papilloma about 0.5 cm. in diameter in each groin. In the small of the back in the region of the third and fourth lumbar vertebrae there were about a dozen brown sessile plaques varying from 1 mm. to 1 cm. in diameter. A similar collection was present over the sacrum.

Central Nervous System.—The cerebral cortex was studded with firm sclerotic masses about 1 cm. in diameter. These could readily be discerned with the finger-tips, but were extremely difficult to see with the naked eye. There was no evidence of hydrocephalus or haemorrhage. The cerebellum was normal. Examination of the pons and medulla showed surprisingly little change considering the decerebrate and acute nature of the man's death.

Urinary System.—The left kidney was congested. Several white pin-point spots were visible on the surface. On cut section a round circumscribed fibrous mass was found at the upper pole 0.5 cm. below the surface. The right kidney showed a similar mass 1.5 cm. in diameter at the lower pole and another small deposit near the pelvis. No abnormalities were noticed in any other organs.

On histological examination of the central nervous system the sclerotic areas showed an increase in glial tissue, with a bizarre cellular structure. Displacement and disorientation of large nerve cells were characteristic (Plate, Fig. 1). The tumour in the left kidney was epithelial in nature and had an adenocarcinomatous appearance (Fig. 2). The other renal tumours were quite different, and consisted of vascular fibrous tissue (Fig. 3).

This patient thus showed many of the classical lesions of epiloia, including adenoma sebaceum, dermal tags and plaques, tuberous sclerosis, and bizarre renal neoplasms. His normal intelligence and the absence of any history of fits were unusual features.

Summary

Sudden death in a young adult is described in which the typical lesions of epiloia were found with recent evidence of normal intelligence and no evidence of fits.

A similar case described by Stewart (1935) did not exhibit adenoma sebaceum or renal neoplasms.

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SIR CECIL WAKELEY AND P. CHILDS: APPENDICITIS

FIG. 1.—Case 40. Mucosa intact; no abnormality in the outer coats apart from vascular dilatation in the subserosa. ($\times 25$.)

FIG. 2.—Case 206. Intense congestion of vessels of the subserosa. Muscle coat oedematous and mucosa intact. ($\times 25$.)

FIG. 3.—Case 17. Showing the earliest changes of an acute inflammatory reaction in subserosa and muscle coats. ($\times 25$.)

FIG. 4.—Complete destruction of the mucosa; all outer coats are heavily infiltrated with polymorphonuclear cells. ($\times 25$.)

FIG. 5.—Section of the distal part of the appendix shows complete replacement of lumen and mucosa by fibro-adipose tissue. ($\times 25$.)

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FIG. 1.—Sclerotic area immediately below surface of cortex. Phosphotungstic acid; haematoxylin. ($\times 300$.)

FIG. 2.—Tumour of left kidney, showing adenocarcinomatous type of growth. H. and E. ($\times 300$.)

FIG. 3.—One of the tumours in the right kidney, showing vascular fibrous tissue. H. and E. ($\times 300$.)