A SPORADIC CASE OF TUBEROUS SCLEROSIS

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Tuberous sclerosis is a rare syndrome, usually familial, characterized by the occurrence of tumours on the surfaces of the lateral ventricles and patches of sclerosis on the surface of the brain. Clinically, progressive mental deterioration, epileptic convulsions, and skin lesions, such as adenoma sebaceum, are usually found. Further manifestations are mentioned below. The rarity of this condition warrants a report being made of a case under my care.

The syndrome was described separately by Hartelegen in 1880 and by Bourneville in 1880-'81, who established it as a morbid entity. It is eponymously associated with him as Bourneville's disease.

Over the years about 400 cases have been reported. It is said[1] that the disease shows no racial proclivity, but that it does not occur in the coloured races. I am informed, however, that a case was diagnosed as such in an Indian infant on X-ray investigation at King Edward VIII Hospital, Durban. The disease is also said to affect males more frequently. In a great number of the recorded cases heredo-familial factors are operative, and in many it appears as a Mendelian dominant.

The child usually seems normal at birth, and the syndrome manifests itself as a rule during infancy or early childhood. The usual milestones of early childhood are achieved rather slowly, and convulsions may occur. In the course of the next few years, the signs of retardation become more evident; and so too do the skin manifestations, which are usually visible before the age of 10 years. At puberty the disease progresses rapidly and, as the child grows older, the mental regression proceeds.

The skin lesions of the face (adenoma sebaceum) become more prominent, and so do the skin lesions on other parts of the body (as in the case to be described).

Temper tantrums are common, and real epileptic manifestations appear; these may be major, minor or Jacksonian in type. Paralysis or paresis is rare.

The mental changes range from complete imbecility to a mild degree of feeblemindedness. In very rare cases the mental powers are only slightly impaired. Some patients show primitive psychotic features, others paranoid hallucinations.

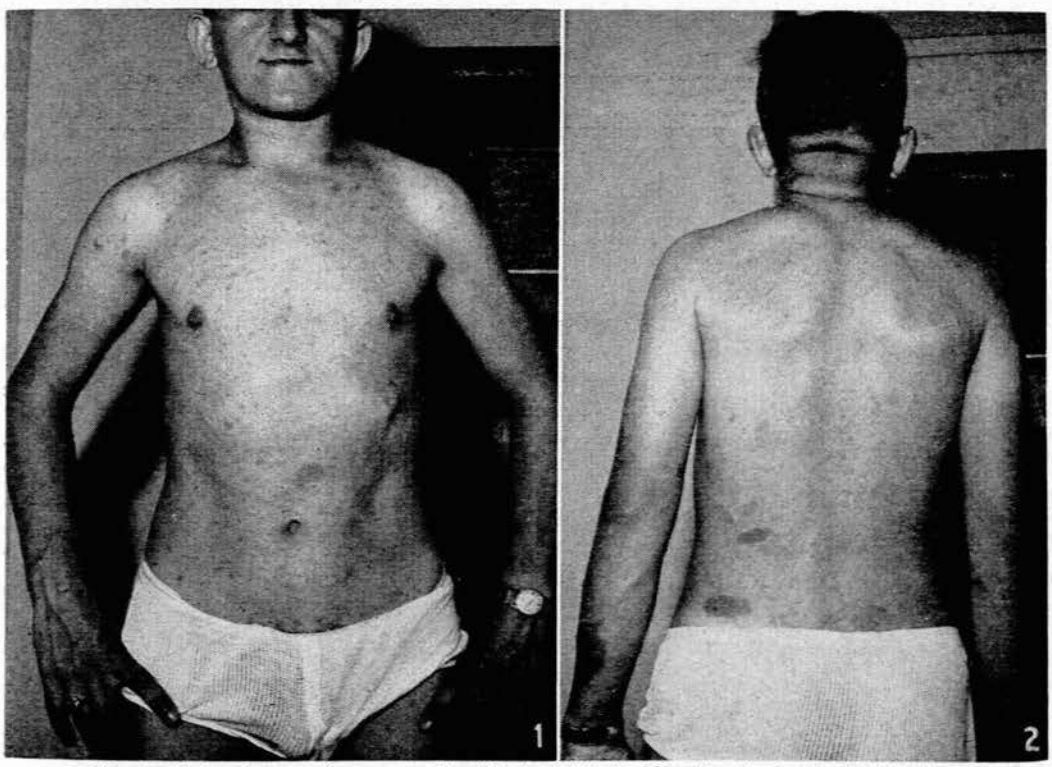
The cutaneous manifestations are an integral part of the symptom complex, and the most common of these is the characteristic facial one in the so-called butterfly distribution.

Although this is usually not the case, the skin lesions sometimes contain neurofibromatous tissue. This link with von Recklinghausen's disease has been described two or three times in the history of the disease. A similar association is noted in the case to be described.

Retinal lesions occur, though rarely. They are usually considered to be a 'phakoma' (congenital tumour) of the retina. In the case to be described the left eye is so disorganized that a clinical diagnosis is quite impossible.

The disease may be associated with other defects and abnormalities in other regions of the body, such as spina bifida, palatal defects, cryptorchia, digital defects, renal-tumour formation, etc.

X-ray changes were first noted by Marcus in 1924. Since that time other X-ray studies have been made and there have been reports of osseous changes and cyst formation in the long bones and the bones of the hands and toes, also of cranial calcification and calcification of brain substance. These X-ray changes are said to occur in about 60% of the patients.



Figs. 1 and 2. Adenoma sebaceum on skin of body and limbs. Note that the face is relatively free.

CASE REPORT

The patient is J.S., a European male, aged 21 years. His parents and two sisters are alive and well. No family history of the disease can be traced.

The patient was a full-term baby and weighed 7¾ lb. at birth. When 10 days old, he developed bronchopneumonia and was very ill. At 3 months he weighed only 6 lb.

He had a tonsillectomy performed at the age of 4 years; at operation there was heart arrest. After the operation the patient did not speak for many months, but in due course speech returned. He spoke, and speaks now, in a voice tone very suggestive of cleft palate (which defect is not present). His mother attributes his present condition to the heart arrest.

He had difficulties in kindergarten and subsequently at school; he was considered a backward child at the age of 10 years.

His facial expression is suggestive of some mental retardation; the lad himself makes very little complaint.

The finger movements are somewhat clumsy. There are morbid temper tantrums, but no true epileptic phenomena are reported. He is blind in the left eye, which is completely disorganized. There are no physical signs of organic nervous disease.

The left testis has not descended. His blood pressure is 120/80 mm.Hg.

His body and limbs are covered with the characteristic skin lesion (Figs. 1 and 2). There are only one or two nodules on the face. A biopsy of one of the nodules from the left forearm showed neurofibromatous tissue.

A Terman-Merrill intelligence test revealed a very low average intelligence.

X-rays of the skull, long bones, hands and feet show no abnormality.

SUMMARY

A sporadic case of tuberous sclerosis is described. No other familial incidence can be traced.

The patient shows the characteristic skin lesion on the body and limbs. There is some intellectual deficit. The occasional association with neurofibromatous changes in the nodules is noted.

I am indebted to Dr. S. Atlas of Durban, who referred the patient to me; and to Drs. B. Kramer and L. E. Wolpert for the X-ray studies.

REFERENCE

1. Bruce, A. N. ed. (1954): *Kinnear Wilson's Neurology*, 2nd ed. London: Butterworth.