

## THE USE OF C.T. SCAN IN THE DIAGNOSIS OF TUBEROUS SCLEROSIS.

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Tuberous sclerosis, one of the phakomatoses, is a genetically determined condition with an autosomal dominant mode of transmission. Bunday and Evans (1969) have demonstrated that up to 86 per cent of cases can arise as the result of new mutations. Nevertheless, the genetic implications of this condition make its early diagnosis imperative. Although the classical triad of features, mental retardation, epilepsy and adenoma sebaceum, are generally accepted as cardinal characteristics, diagnosis can still be difficult to establish as the individual signs of this triad are not always present.

First, not all individuals with tuberous sclerosis are mentally retarded. Borberg (1951) in Denmark and Nevin and Pearce (1968) in the United Kingdom have carried out two population studies of note. No fewer than 11 of Borberg's 37 cases and 6 of Nevin's 18 cases were of normal intelligence.

Second, epilepsy may be absent. In the study of Lagos and Gomez (1967) in the Mayo Clinic, only 69 per cent of the retarded group had seizures.

As regards adenoma sebaceum, Bjornberg (1961) considers this as being always part of the tuberous sclerosis complex. Bunday and Evans (1969), however, are of the opinion that shagreen patches, subungual fibromata and possibly white naevi indicate the presence of the gene for tuberous sclerosis in the absence of adenoma sebaceum. They believe that when adenoma sebaceum is not present, the diagnosis of tuberous sclerosis may be made solely on the finding of a retinal phakoma.

The following two cases illustrate the difficulties of diagnosis:

J. W. was born on 30.3.1964. His mother had pyelitis at 32 weeks gestation. She went into spontaneous but prolonged labour. Early signs of maternal and foetal distress necessitated low forceps delivery. The child developed infantile spasms at the age of six months with apparent arrest of mental development. Serial E.E.G.'s showed abnormalities and at the age of 1 year 3 months, the E.E.G. showed hypsarrhythmia. The patient was admitted at the age of 2 years 4 months with a diagnosis of severe subnormality, epilepsy and hypotonia. Fits stopped at the age of 3 years. At the age of 13 years 3 months, the E.E.G. showed a pattern which was interpreted as potentially epileptogenic with mild diffuse abnormalities. The course was then uneventful until the age of 16 years when the patient started deteriorating. He became unable to walk, and had to be confined to bed. He was also unable to take feeds. He was then seen by a neurologist. His freckled face could not be interpreted as adenoma sebaceum. The patient was admitted to Great Ormond Street where a scan revealed tuberous sclerosis. The patient died of bronchopneumonia three weeks later. Post mortem examination showed several typical plaques in the cerebrum up to 3 cm., particularly over the right temporal lobe and left frontal region.

At the time of the patient's birth his parents were 17 years and 18 years of age. They had two normal children subsequently. They were urged to attend for dermatological examinations to establish whether or not they were carriers, but they have not availed themselves of this.

M. M. was born on 9.8.1959. Her mother was epileptic. Her mother's sister had an epileptic son. Her paternal grandfather had epilepsy. The parents were of Irish origin. The patient's mother had toxæmia of pregnancy for which she was in hospital for two months. Forceps delivery was carried out at 43 weeks. Birth weight was 8lbs. 2oz. The immediate post-natal period was uneventful. Fits began in the third month. Her mother also noted

backwardness. Severe subnormality was diagnosed at the Hospital for Sick Children, Great Ormond Street at the age of 3 years. The patient was subsequently admitted to long-stay hospitals for the mentally handicapped. Tuberous sclerosis was suspected because of microaneurysms of the retina, and various skin lesions, namely plaques, on the face, white shagreen patches on the back over the spine and a patch of leucoderma on the abdomen. The pathologist's report on the skin biopsy, however, read: "non-specific and of no diagnostic value". The patient was discharged at the age of 14 years against medical advice. She died at the age of 21 years. Post mortem examination confirmed a diagnosis of tuberous sclerosis.

## DISCUSSION

In both the illustrated cases, a definitive diagnosis could not be made solely on the classical triad. Indeed, Barakat and Cochran (1978) believe that computerized tomography is the most reliable approach to an early diagnosis of tuberous sclerosis. Furthermore, they recommend that a complete investigation of immediate family members of affected cases, including computerized tomography, would demonstrate a higher familial incidence and help clarify the mode of transmission.

Lorusso, Clochiatti and De Luca (1979) made a diagnosis of tuberous sclerosis in an 8 months old boy with generations of parent-to-child genetic transmission by means of computerized tomodensimetry. Logos and Gomez (1967) are of the view that the diagnosis tuberous sclerosis can be confirmed when a patient has roentgenologic evidence of circumscribed intracranial calcifications. These can be searched for by pneumoencephalography or computerized tomography.

It is therefore recommended that in all cases of suspected tuberous sclerosis not presenting with the classical triad *in toto*, a C.T. be performed.

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