TUBEROUS SCLEROSIS : CASE RECORD AND DISCUSSION

VICTOR BERMAN, M.B., CH.B. (CAPE TOWN), D.M.R. (LOND.), *Radiologist in charge, Transvaal Memorial Hospital for Children, Johannesburg*, and EDWIN MYER, M.B., B.CH. (RAND), *Johannesburg*

In this article we report a case of the relatively rare condition known as tuberous sclerosis, and add a brief description of the salient features of published cases with particular reference to radiological findings.

CASE REPORT

A European boy aged 11 years 9 months was brought to the Children's Hospital because of fits which he had been having since the age of 18 months. These fits consist of an aura with frequent repeating of words and rapid clenching of his hands. There is no cyanosis or crying. He usually staggers or falls to the left and there is a loss of consciousness lasting 3-10 minutes. He has had as many as 17 of these attacks in a day and at other times 1 or 2 per week. Between these attacks the boy leads a reasonably normal life. He is fond of music and reads and draws well. He attends a special school, being mentally retarded.

Family History. The mother was ill during her pregnancy but cannot say what was wrong. Delivery was with forceps but the child was well at birth. Two siblings are normal. There is no history of epilepsy in the family. The mother has a similar skin condition on her face to that of the boy (Fig. 1).

Examination. The child appears to be mentally dull and retarded, clenching and rubbing his hands all the time. As he was being admitted he jumped from a 10-foot balcony, fracturing a cuboid; soon afterwards he bit a thermometer. Adenoma sebaceum is present on the face (Fig. 1). Nothing of note found in the central nervous, cardiovascular and respiratory system, the ear, nose and throat, the abdomen and skeleton.

Special Investigations

Cerebrospinal Fluid. No cells, chemically normal, protein normal, Wassermann reaction negative.

Electro-encephalogram. Random delta theta frequencies and dominating all leads. Minimal amount of 8 c/s alpha activity was recorded at low amplitude over the central and post-central areas. Frequent short episodes of typical slow waves and spikes were recorded at high amplitude in all leads over both hemispheres. This abnormal EEG is regarded as consistent with the diagnosis of tuberous sclerosis.

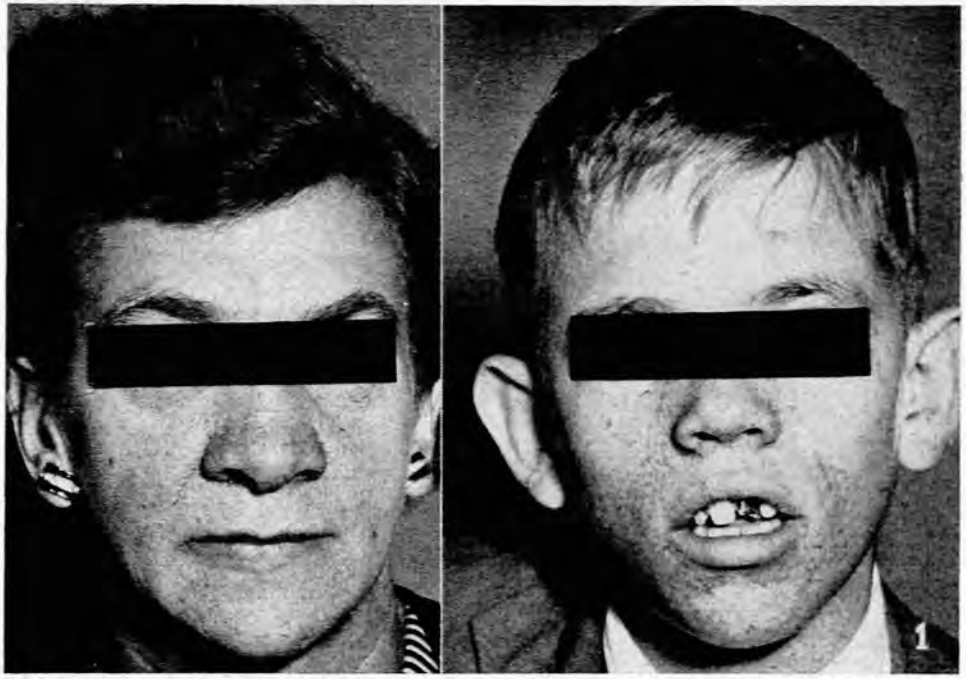

Fig. 1. Picture of the patient and his mother. Note the adenoma sebaceum of the face, more marked in the boy.

Radiological Examination

Chest normal.

Intravenous Pyelogram. The kidneys excreted the dye normally. The collecting system on the right side was normal. On the left side there was some thinning of the pelvic-calycal system suggesting some pressure from the kidney parenchyma. No calcification could be seen in the kidneys.

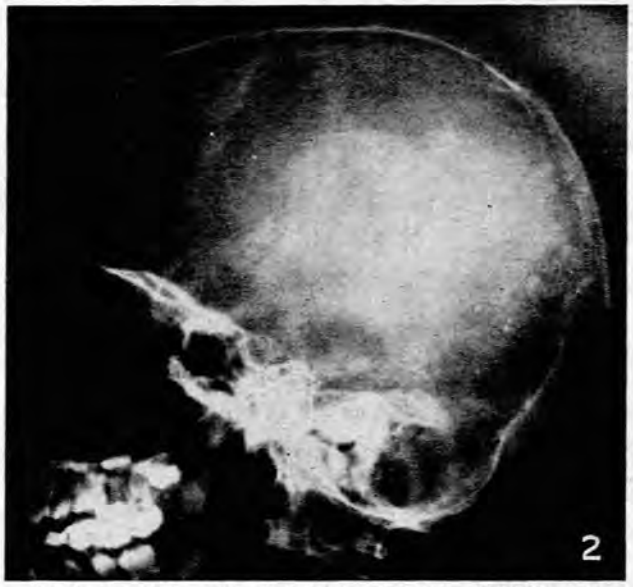

Fig. 2. Lateral view of the skull showing the intracranial calcification.

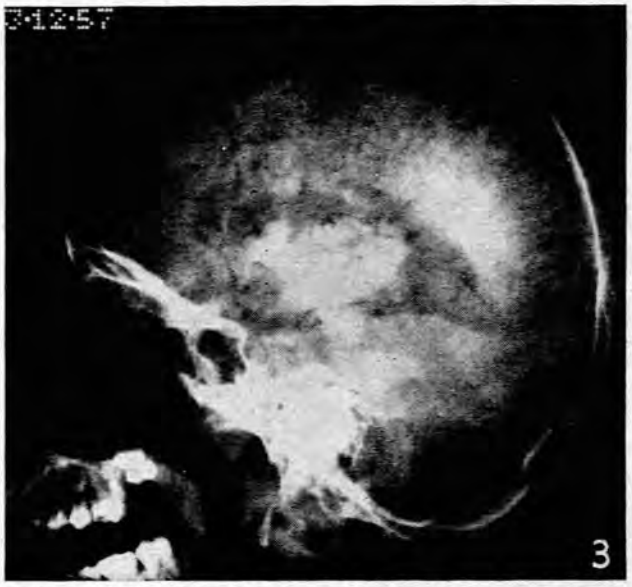

Fig. 3. Lateral view of the air-study showing the close association of the calcification with the lateral ventricles.

Skull. The straight films of the skull show some calcification just anterior to the coronal sutures in the lateral view (Fig. 2) and close to the mid-line in the antero-posterior projection. Air studies of the ventricular system (Figs. 3 and 4) show that this calcification is adjacent to the infero-lateral walls of the lateral ventricles. The characteristic 'candle guttering' described in the literature was not well shown in this series of films.

Hands. A small cystic area with clear-cut margins is present in the tuft of the terminal phalanx of the left third finger; similar but less clearly defined, cystic areas can be seen in the terminal phalanges of the right index and fourth fingers (Fig. 5). The wavy periosteal reaction in the phalanges described in the literature as being characteristic of the condition is not seen in these films.

DISCUSSION

The condition of tuberous sclerosis was described as far back as 1880 by Baurneville. It was so named because of the potato-like lesions in the brain which form the basis of its pathology. These lesions are smooth, white and fairly hard.

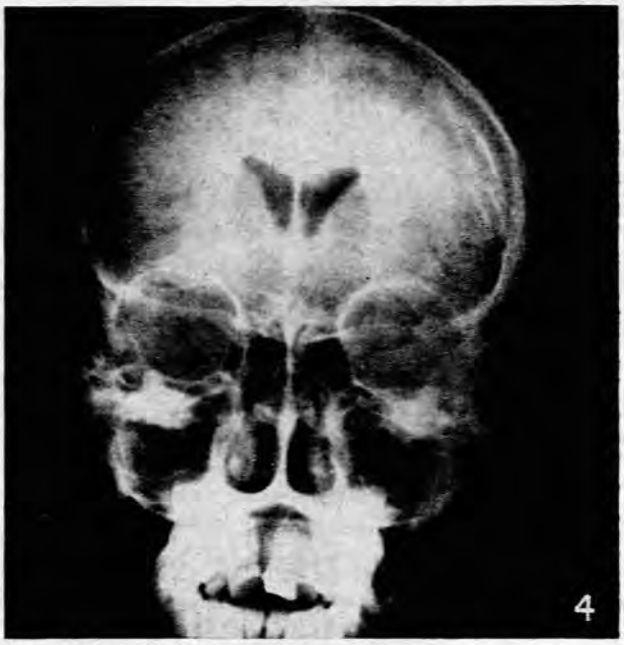

Fig. 4. Antero-posterior view of the air-study showing the relationship of the calcification to the lateral ventricles. This is seen far more clearly in the original X-ray film.

Radiologically, tuberous sclerosis is most commonly associated with calcification in the brain, usually in the region of the basal ganglia. Of a series of 43 cases Holt and Dickerson[1] found this feature in 50%. The lesions may at times be seen to project into the ventricles and when this is shown with air-studies Ross and Dickerson[2] regard it as pathognemonic.

Clinically, the patient with tuberous sclerosis presents himself with facial adenoma sebaceum, mental retardation and epilepsy or convulsions, but not all of these features need be present at the same time. The mother of our patient has facial adenoma sebaceum but is mentally normal and radiologically negative.

Retinal tumours (phakomas) are said to be fairly common in this condition, and also renal tumours, which usually consist of a proliferation of mesothelial elements including muscle, fat, fibrous tissue and vessels. Cysts have been seen to occur in most of the viscera, resulting in a wide variety of clinical manifestations.

Our case shows osseous lesions in the fingers which are seen far less frequently than intracranial calcification. Holt and Dickerson,[1] in their admirable paper on the osseous lesions of tuberous sclerosis, state that 66% of 30 patients who had X-rays taken of their hands and feet showed local cystic lesions in the phalanges with or without associated periosteal formation of new bone in the metacarpals or metatarsals. This periosteal process presents a 'wavy' appearance, they say. Ackermann[3] has reported cases of tuberous sclerosis with very striking osseous manifestations, but our case does not show these changes.

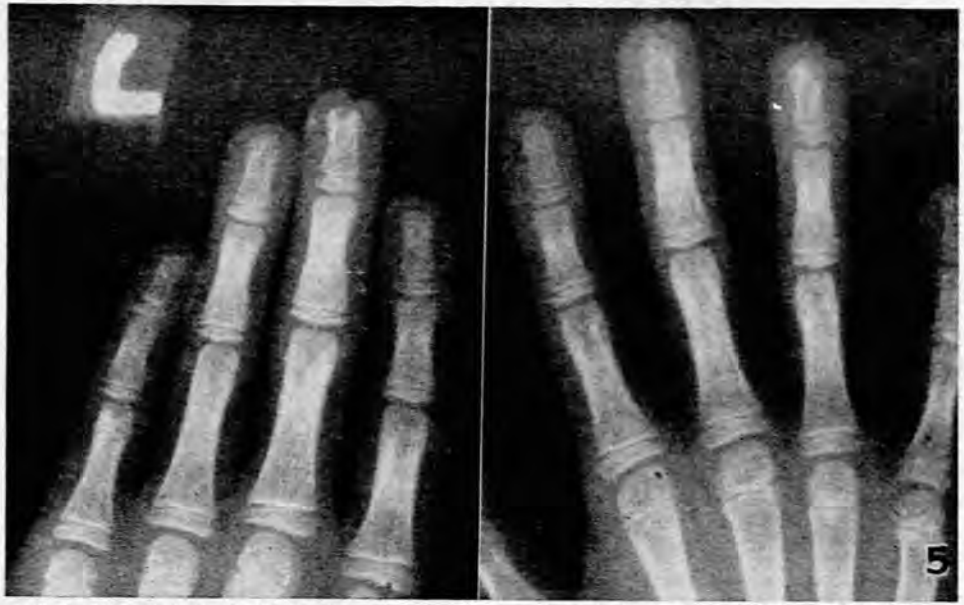

Fig. 5. Film of the hands showing the cystic lesions in some of the phalanges. The one in the terminal phalanx of the left 3rd finger is particularly well seen.

SUMMARY

A case of tuberous sclerosis in a boy of 11 years is described.

The case presented with adenoma sebaceum of the face associated with mental retardation and fits.

The radiological features included intracranial calcification adjacent to the lateral ventricles. Osseous manifestations were found in the phalanges of the hands.

Some of the main features of the condition are described.

We wish to thank Dr. K. F. Mills, Superintendent of the Johannesburg Hospital, and Dr. J. L. Parnell for permission to publish this case. We also thank Mrs. Hill, of the General Hospital X-ray department and Mr. Schevitz, of the Medical School, for assisting with the reproductions.

We should like to add that this case was first seen in the outpatients department of the Children's Hospital by Dr. Johann Theunissen who, but for his untimely death, would have been associated with this publication.

REFERENCES

1. Holt, J. F. and Dickerson, W. W. (1952): Radiology, **58**, 1.
2. Ross, A. T. and Dickerson, W. W. (1943): Arch. Neurol. Psychiat. (Chicago), **50**, 233.
3. Ackermann, A. J. (1944): Amer. J. Roentgenol, **51**, 315.