TUBEROUS SCLEROSIS

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Tuberous sclerosis is a rare congenital disease of unknown aetiology. It is also known as Epiloia or Bourneville's disease. The disease is characterised by mental retardation, epileptic convulsions and adenoma sebaceum.

We had the opportunity of studying three such cases during the last six months These three cases are presented and the subject is reviewed.

CASE REPORT:

Case No. 1:

Rajan, male patient aged 20 years was admitted for epileptic convulsions. Patient could not give relevant history as he was mentally retarded. On examination, he had adenoma sebaceum (plate 1). He was fairly built and had multiple scars on the extremities probably due to trauma, following convulsions. His left leg was short due to deformity at the ankle joint. Patient had idiotic look and he could count upto ten digits only. He looked to be confused and memory for past events was impaired. He was disoriented in time. Otherwise neurological examination did not reveal anything abnormal. Other systems were normal.

Investigations:

Hb : 10 gms.%
WBC count: 8,000/cmm.
Urine : N.A.D.
Blood VDRL : Negative
CSF : Normal
CSF-WR : Negative

X-ray chest: N. A. D.

X-ray hand: Metacarpals showed cystic changes and irregular periosteal thickening.

X-ray ankle: (Left) Gross talipus with irregular posterior aspect of calceneum.

X-ray skull: Multiple dense sclerotic patches seen all over (Plate 2).

Air-encephalogram: Tumour like mass was seen protruding into the lateral ventricle (Plate 3).

X-ray lumboscral region: Flame shaped opacities seen.

I. V. P. : Abnormal shape of left kidney.

Renal angiography: Abnormal shape of left kidney otherwise normal (Plate 4).

Funduscopy: Multiple phakomata were seen in both eyes.

EKG : within normal limits.

Case No. 2:

A male patient 20 years of age attended the skin and V.D. out-patient department for severe itching. He had scabies which was treated. He was found to be mentally retarded and was formerly admitted in the Mental Hospital. His relative revealed that he used to get epileptic fits from the age of 2½ years. On clinical examination he was fairly well

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built. He had multiple scars on his face following epileptic fits which could not be controlled with 3 grs. of phenobarbitone and 600 mg. of dilantin. There were extensive hyperpigmented papular lesions on the molar region.

Skin over the sacrum showed two thickened areas of shagreen patches. He had vitiliginous patches over the lower extremity. There was no evidence of subungual fibromas. No other members of the family were affected.

Investigations:

X-ray of hands showed cystic areas in metacarpals.

X-ray skull showed obliteration of diploic space and few opaque patches were seen.

X-ray of dorsolumbar spine did not reveal any abnormality.

Funduscopy did not show any evidence of phakomatoses.

EKG was within normal limits.

Skin biopsy (from the face) showed hyperkeratosis and slight acanthosis but there was no increased number of subaceous glands or increased vasculature.

Case No. 3:

A female patient 21 years old came first to the skin and V.D. department for adenoma sibaceum, vitiliginous patches and cafe' au lait pigmented spots on the chest and back. She was mentally retarded and was educated only upto 4th standard. She was suffering from epilepsy since early childhood and was controlled with phenobarbitone.

Investigations:

Her X-ray of hands did show cystic changes and irregular periosteal thickening of the metacarpals. X-ray skull revealed thickening of diploic space.

Dermatoglyphics:

The finger print patterns in all our three cases did not show any significant dermal pattern. An interesting thing noted was the presence of loop-fibular pattern (loop facing the fibular side of the foot) on the hallucal area of both the feet in all the three cases. This pattern is very unusual in the control group studied and its diagnostic significance would be better evaluated in a lage series.

Discussions:

Tuberous sclerosis is genetically inherited disease and is transmitted as a single antosomal dominant gene and has variable expression.

This rare disease usually manifests between the age group of 5 and 17 years (average 9 years) and 75% die before reaching the age of 20. There are reports where patients have survived longer than that. All three cases of ours were beyond 20 years of age. It was emphasised by Brain that the disease is seen only in white race but recently Cosnett et al have described the disease in three Zulu patients from Africa.

Though mental retardations are present invariably in all cases recently a few cases are reported where this essential feature of the disease was absent (Bhardwaja et al). In some instances both epilepsy and mental retardation were absent. All three of our cases had significant mental retardation and epileptic fits from childhood. One of them had convulsions from $2\frac{1}{2}$ years of age and was resistant to high doses of anticonvulsant drugs like phenobarbitone and dilantin.

Adenoma sebaceum – symmetrically distributed fresh colored papules was described in 1885. This skin lesion often brings the patient to the dermatologist first. The lesion is so characteristic that it is difficult to miss it. Shagreen patches, skin tags, subungual fibromata, retinal

phakomas, (thus included under phakomatoses) and very rarely cardiac arrhythmias like ectopic beats and conduction defects (Cosnett) are reported in the literature. Rhabdomyoma of heart (congenital nodular glycogenic infiltration of myocardium) is often associated with this disease;. tumours in thyroid, thymus, breast and duodenum are also described.

Renal masses are present in 50 – 80% of cases and often they are multiple and bilateral. The tumour invariably turns out to be angiomyolipoma. In case No. 1 left kidney was abnormal in shape but no definite evidence regarding the nature of the mass could be demonstrated on angiogram. A case is reported (Anderson) where patient was maintained on haemodialysis for 21 months for chronic renal failure.

Orthopaedic deformities of various nature are reported (Smith et al). In their series of 32 cases, they found 7 were toe walkers, 2 had scoliosis and 2 had talipus equinovarus deformities. Hip dislocation, contractures are also seen in some cases.

For confirmation of clinical diagnosis, radiological investigations are of immense value. They are as follows:

1. a) Skull: Intracranial calcification around basal ganglia is seen in 50% of cases. Rarely calcification (7-15%) is seen in cerebellar region, medulla and cord.

   b) Sclerotic areas in calvarium with obliteration of marrow spaces in diploe are seen in about 40% of cases.

   c) Air encephalogram shows candle guttering appearance due to protrusion of tuberous patches.

2. Hand and feet: Cystic areas with smeared borders are seen in metacarpals and metatarsals in 60% of cases. Periosteal new bone formation with irregular wavy margins is noticed.

3. Lumbar vertebrae and Pelvis: Osteoblastic (sclerotic) areas are seen in lumbar vertebrae particularly in the region of pedicles. They are round or oval in shape and measure from few mms. to few cms. Flame shaped sclerotic lesions are observed sometimes as in cases of Paget's disease.

4. Humerous and Femur: Thickening of cortex of diaphyses is seen.

5. Chest: Honey-combed appearance of lungs is often noticed, resembling that seen in Hammann-Rich syndrome or Sarcoidosis. Diffuse interstitial fibrosis and emphysema is also observed. Rarely still, pleural effusion may be present (Broughton).

6. I. V. Pyelography: Enlarged kidney shadows and or appearance resembling polycystic kidneys is sometimes seen but lesion may turn out to be angiomyolipoma or adenosarcoma. However, they are frequently asymptomatic.

7. Renal angiography might reveal presence of angiomatous malformation.

Summary

Three cases of tuberous sclerosis are presented and various methods of investigations are described. The subject is discussed in short.

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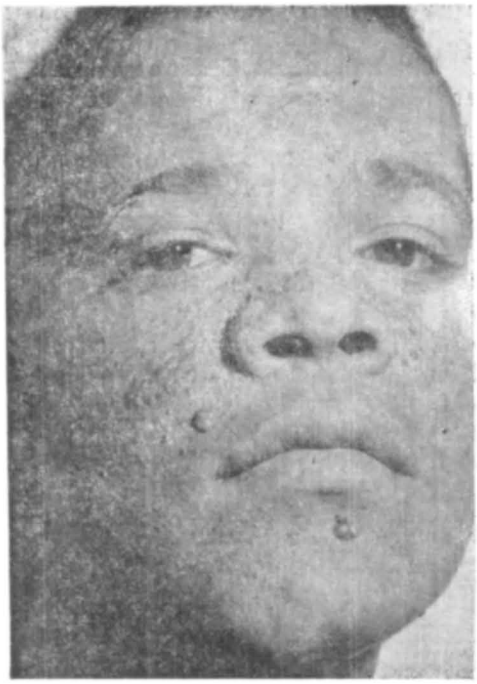

Plate 1
Adenoma Sebaceum

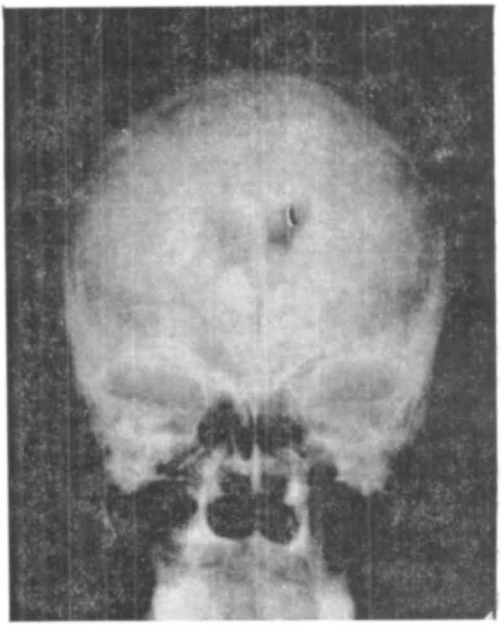

Plate 3
Air-encephalogram—tumour like mass seen protruding into lat. ventricle

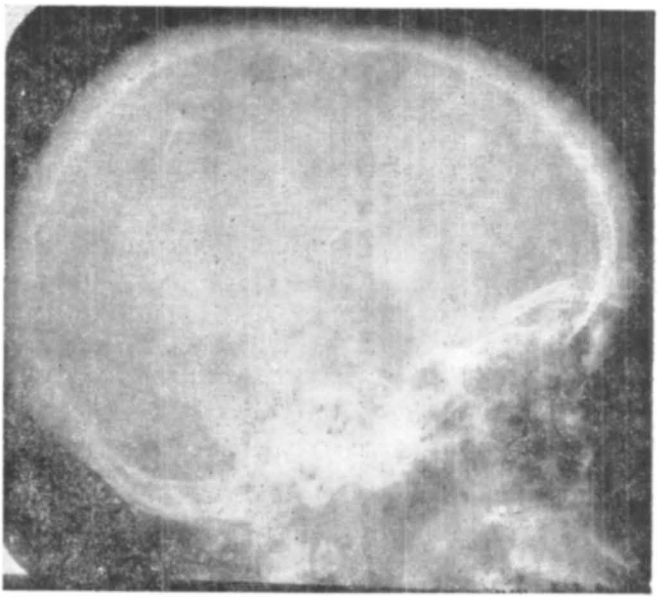

Plate 2
X-ray skull—multiple dense sclerotic patches

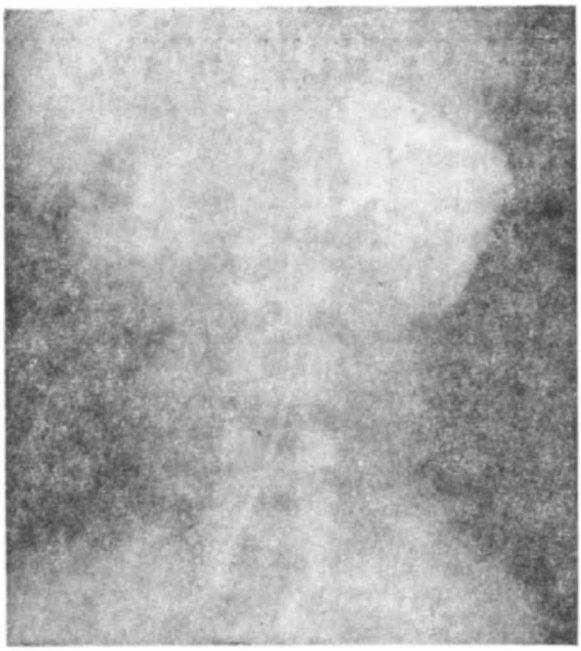

Plate 4
Renal Angiography—Abnormal shape of left kidney

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