RENAL ANGIOMYOLIPOMA IN A FAMILY WITH TUBEROUS SCLEROSIS

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SYNOPSIS

This case report describes a family with forme- fruste tuberous sclerosis and renal angiomyolipoma. The clinical manifestations of tuberous sclerosis are discussed.

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INTRODUCTION

Tuberous sclerosis was first described as a pathological entity at the end of the nineteenth century by Bourneville. He named it after the macroscopic appearance of cerebral lesions. However, it was von Recklinghausen who published the first pathological discription in the year 1863. The disease is hercditary and familial in most cases. This disease usually leads to death within the first years of life but many forms are less severe and have a later development of visceral lesions. These lesions are most frequently renal.

We describe a family with tuberous sclerosis. The renal manifestions are highlighted in the discussion.

CASE REPORT

NSH, a 28 year old Chinese lady presented in Oct '86 with pain localised to the right loin of 4 days duration. The pain was constant and described as pulling in nature. There was no history of hematuria, dysuria or frequency of micturition.

The patient had a history of fits from the age of 7 years, which lasted for several years during which she was on medication. However, she absconded follow-up 15 years ago, and has since remained well.

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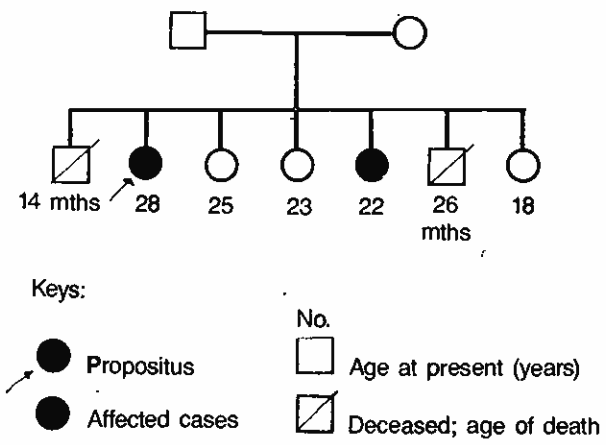
In the family history, it was noted that 2 sibs died at the ages of 14 months and 2 years. Only the latter sib had a history of fits. The family tree is as follows (fig 1):

Fig 1: Family history



On examination, the patient was afebrile. Pulse rate 78/min and blood pressure 110/80 mmHg. There was no pallor. Over both cheeks, adenoma sebaceum were present. There were hypopigmented macules and shagreen patches over her back. Sub-ungal fibromata were seen on both hands.

Fundoscopy revealed phakoma of right eye. In the abdomen, both kidneys were palpable. There were no bruits heard. There were no other abnormal physical findings.

Urinalysis showed microscopic hematuria of 10 rbcs per hpf. Skull X-rays were normal. CT scan of the abdomen (see fig 1) showed grossly enlarged kidneys bilaterally. There was a mixture of fat and solid masses within it. The findings were consistent with angiomyolipoma. Gastroscopy showed giant gastric polyps. Histology was consistent with hamartomatous lesions of gastric mucosa. Intracranial calcifications were seen on CT scan of the cranium (see fig 2). The surviving

Fig 1 Bilateral renal angiomyolipoma on CT scan

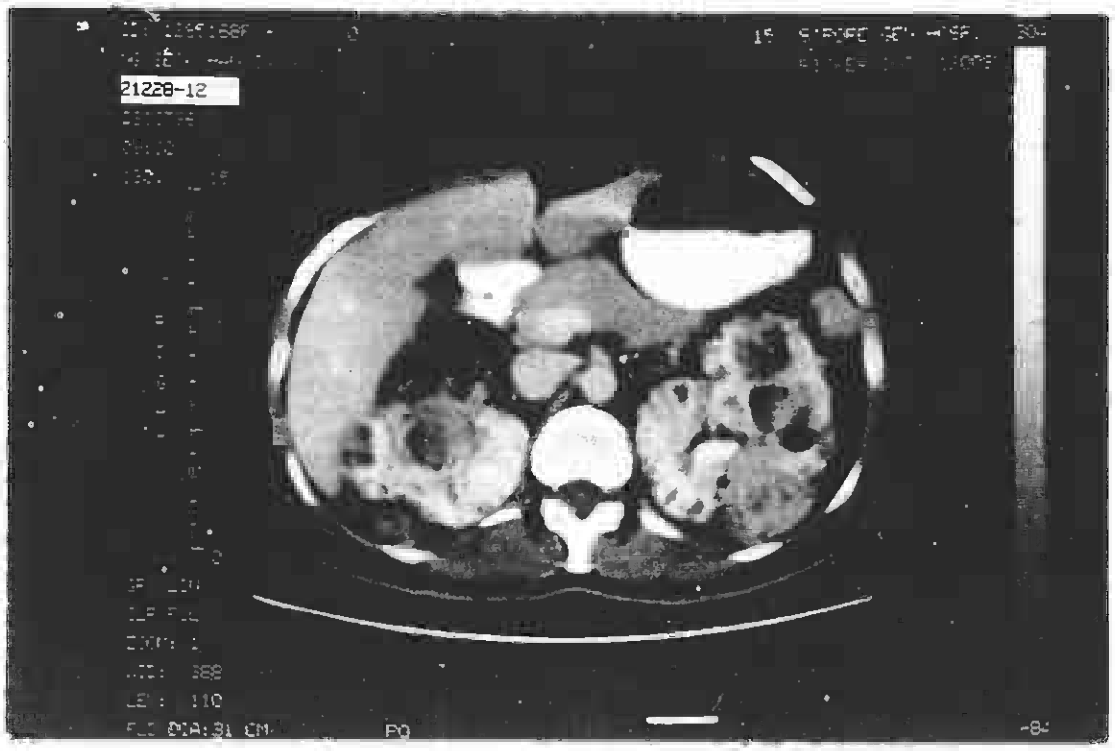
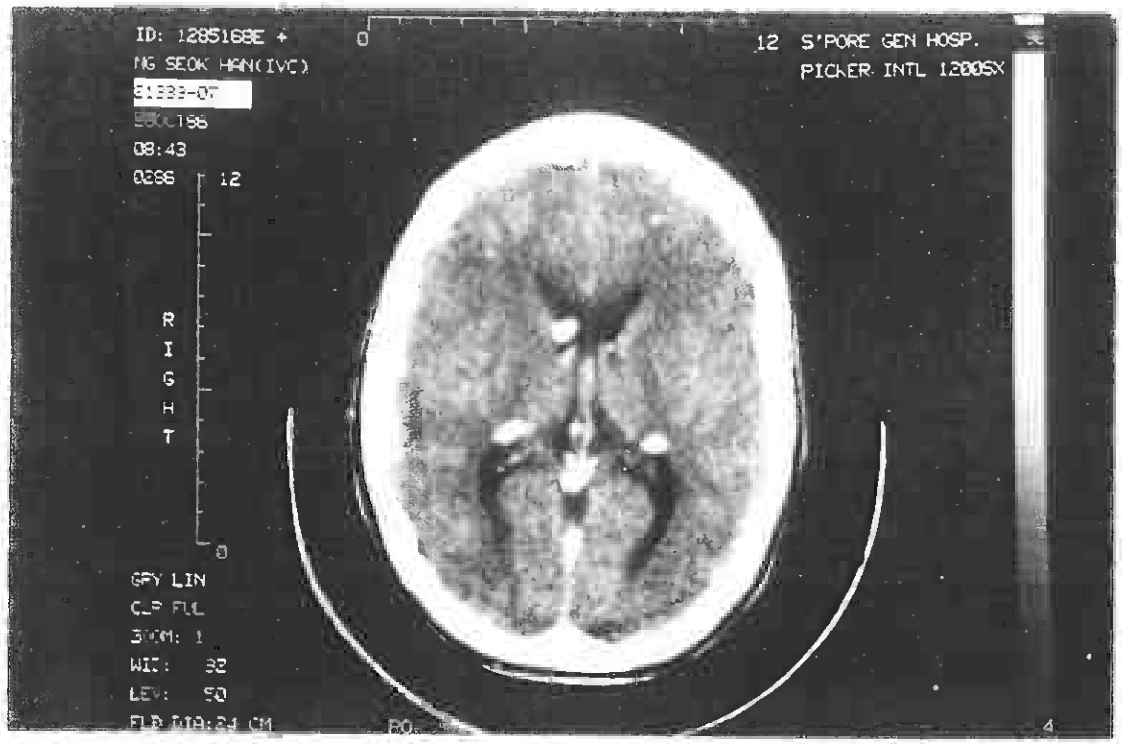


Fig 2 Intracranial calcifications on CT scan



members of this family were examined and tuberous sclerosis was diagnosed in her father and one of her sisters. There was no history of fits. Clinically, both patients had bilaterally enlarged kidneys. CT scan findings were again consistent with angiomyolipoma.

DISCUSSION

Von Recklinghausen (1) first described Tuberous sclerosis (TS) at an autopsy which showed sclerotic lesions of the brain and multiple tumors of the heart. In 1942. Moolten(2) recognized that it was "a congenital developmental anomaly characterised by the occurence of tumor-like malformation in various organs. Involving those of ectodermal origin, brain, skin, retina(3), but also those of mesodermal origin notable the heart and kidneys."

TS is an inherted condition characterised by a clinical triad of epilepsy, mental retardation and adenoma sebaceum. It is accepted that a forme fruste exists. These patients may have only one feature of the triad.

Epilepsy(4) is present, at one time or another, in 90% of the patients. It may feature predominantly or may be a one time occurence. In 70% of the cases, skull x-rays will show intra-cranial calcifications. The sclerotic glial nodules are often found at the basal ganglia and paraventicular regions.

About two-thirds of cases with TS are mentally retarded(4,5). All the mentally retarded patients will have a history of seizures. In contrast, only 70% of the patients with average intelligence will have seizures. In this reported family, the three affected were of average intelligence. Only the index case had history of fits.

"Adenoma sebaceum" are angiofibromas which are seen over the cheeks and the nose. They are often the feature which alerts the clinician to the diagnosis of TS. Of the skin lesions, adenoma sebaceum are the most prominent. Others(6) include shagreen patch, hyperpigmented or achromatic patches, cafe au lait spots and periungal or subungal fibromas.

Renal involvement (4,7) is present in 40% to 80% of the patients with TS. Of these, angiomyolipoma(8,9) is the most common. Other renal manifestations include polycystic kidneys and renal cell carcinoma. They may present with flank pain, episodic hematuria or flank masses.

The clinical differentiation between angiomyofibroma and polycystic kidnerys is difficult. Although proteinuria is more common in polycystic kidnerys, it can also be found in angiomyolipomas. In angiomyolipoma, the onset of symptoms tends to be earlier. Most cases develop renal symptoms by the age of 25. In contrast, polycystic kidneys(10) are symptomatic from age 35 to 50 years. The incidence of renal failure is low in TS. Thsi has been attibuted to early death from repeated seizures. However, with more effective anticonvulsants and neurosurgical excision of epileptogenic foci, renal failure may become more common in TS.

The intra-venous pyelogram in both conditions are similar. Bilateral renal enlargement and distortion of the renal calyceal system is seen in both conditions. Sometimes, lucent areas scattered throughout the renal parenchyma can be seen on tomograms at the time of intra-venous pyelogram. These lucent areas correspond to the adipose tissue in angiomyolipoma. At ultrasound and CT scan, the two conditions can be differentiated. One will show multiple cystic lesions. In the other, solid masses with the density of fat and muscle are seen. The hypervascularity with abnormal vessel distribution can be demonstrated on renal angiogram. The main complication that may occur in patients with angiomyolipoma is retro-peritoneal haemorrhage. When this occurs, nephrectomy is often necessary.

In addition to having angiomyolipoma, this patient also had phakoma of the eyes and gastric harmatomatous polyps. These developmental anomalies are often seen in TS.

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