ACUTE PRESENTATION OF TUBEROUS SCLEROSIS: CASE REPORT

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ABSTRACT

Incomplete form of tuberous sclerosis (TS) may present with acute complications such as haematuria, retroperitoneal haemorrhage or pneumothorax. Such cases may pose diagnostic difficulty. A patient with incomplete form of TS without any cerebral impairment who presented as an acute surgical abdomen is reported. The diagnostic criteria of TS are reviewed. Visceral manifestations of TS including acute complications are discussed. The importance of recognising such presentations is stressed.

Keywords: tuberous sclerosis, incomplete form, visceral manifestations, acute presentations.

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INTRODUCTION

Tuberous sclerosis (TS) is a rare heredofamilial disease characterised by a variety of haematomatous lesions in the brain, skin, retina and viscera. Clinical diagnosis is easy when the patient presents with classical triad of seizures, mental retardation and adenoma sebaceum[1]. However, from time to time the incomplete form (forme fruste) without cerebral impairment or facial eruptions may present. When such patients present with acute visceral complications such as pneumothorax, haematuria or acute abdominal pain[2], serious mistakes in diagnosis may occur if the possibility of TS is not considered. A case of incomplete form of TS who presented as an acute surgical abdomen is reported. Visceral manifestations of TS including acute visceral complications are discussed. The importance of recognising such presentations is stressed.

CASE REPORT

A 24-year-old Chinese man was admitted to the surgical ward with a history of sudden onset of abdominal pain. His past history was unremarkable.

On physical examination he was pale with a pulse rate of 116/minute and BP of 120/60 mmHg. His intellect was normal. The abdomen was tense and tender especially of the left side. Needle aspiration of the abdomen revealed blood. His haemoglobin was 8.0 gm/L.

A diagnosis of intra-abdominal bleeding was made and an emergency laparotomy was performed. At laparotomy he was found to have a large retroperitoneal haematoma on the left side and he was bleeding from the left kidney. Both kidneys were enlarged with multiple mass lesions. This was misinterpreted as polycystic disease of the kidneys. Left nephrectomy was done. During the post-operative period which was uneventful, the following investigations were done:

(a) Intravenous Urography
The right kidney was enlarged with stretching and deformity of the pelvicalyceal system indicating the presence of multiple mass lesions (Fig 1).

(b) Ultrasound examination of the kidney
The right kidney was enlarged with multiple echogenic mass lesions. The appearance was consistent with angiomyolipomas (Fig 2).

(c) X-ray of skull
Sclerotic islands were seen in the calvarium.

(d) X-ray of pelvis
Sclerotic areas were noted in the iliac bones.

(e) X-ray of hands
Cortical cysts were seen in the phalanges.

(f) Chest x-ray - Normal

Fig 1 - Intravenous urography. The right kidney is enlarged with stretching and deformity of the pelvicalyceal system. The left kidney has been resected. Note the sclerotic areas in the pelvic bone.

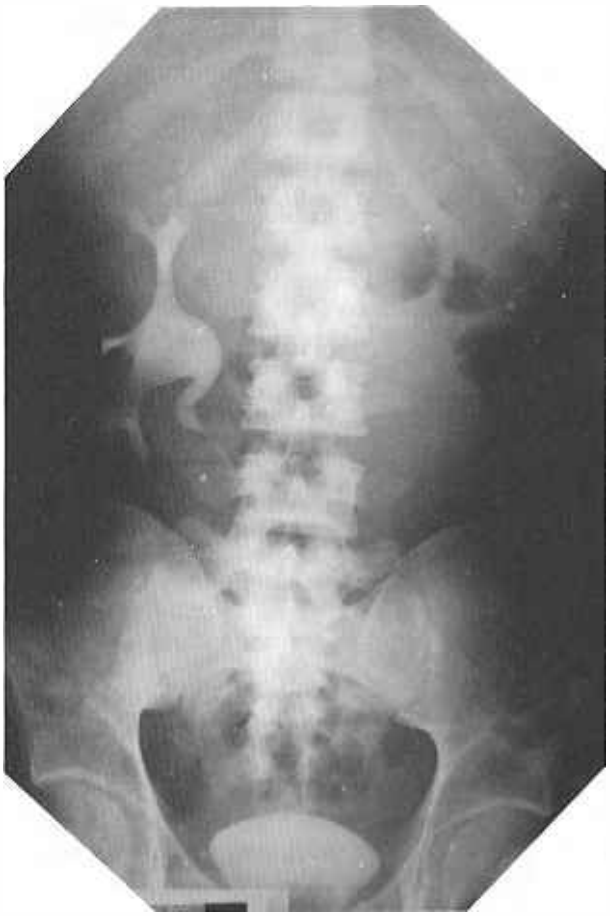

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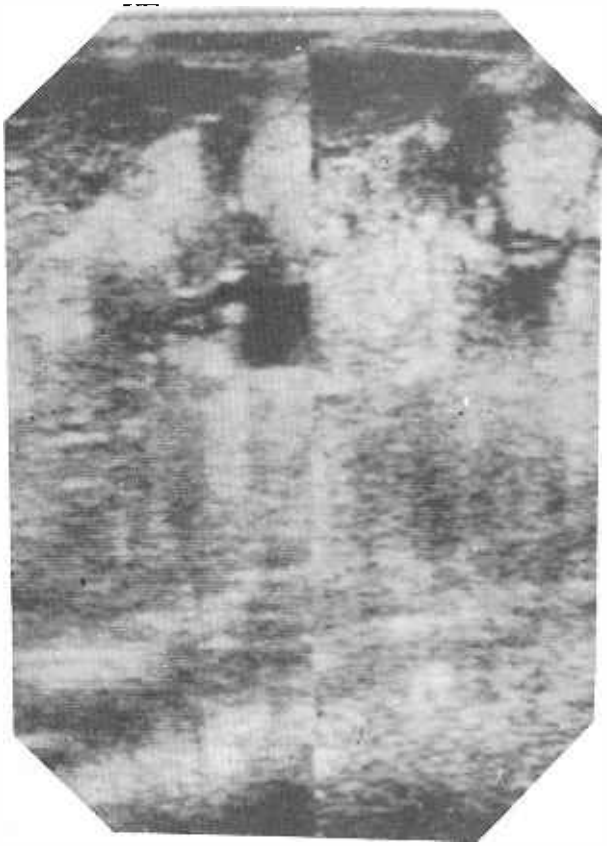
Fig 2 - Ultrasound of the right kidney. Multiple echogenic tumours are seen. This appearance is consistent with angiomyolipomata.



On close examination the patient was found to have the following skin lesions: adenoma sebaceum on the face, shagreen patch on the right loin, subungual fibromata and disseminated small hypopigmented lesions.

A diagnosis of TS was made. Histological examination of the resected kidney revealed multiple angiomyolipomas. The patient was discharged well after two weeks and was followed up at the General Hospital, Kuala Lumpur.

DISCUSSION

TS has a wide variability in its presentation and affects many body systems. The criteria for diagnosis give some indication of the variability of this genetic disorder. Criteria for a definite diagnosis of TS include one or more of the conditions listed in Table I, preferably in multiple forms[3]. Criteria for a presumptive diagnosis of TS are listed in Table II[3].

Table I - Criteria for definitive diagnosis of tuberous sclerosis

Facial angiofibromas
Ungual fibroma
Retinal haematoma
Cortical tuber
Subependymal glial nodule
Renal angiomyolipomas

Mental deficiency (71%) with seizures (78%) is the most common clinical presentation[4]. Adenoma sebaceum of the face is the most readily recognised sign of TS but only 47% of the patients have this sign[5]. Other cutaneous lesions include: hypomelanotic macules (86%), subungual fibromas (20%) and shagreen patches (19%). The patient presented here had all the cutaneous lesions mentioned above.

Table II - Criteria for presumptive diagnosis of tuberous sclerosis

Hypomelanotic macules
Shagreen patches
Peripapillary retinal haematoma
Gingival fibromas
Dental enamel pits
Single renal angiomyolipoma
Multicystic kidneys
Cardiac rhabdomyoma
Pulmonary lymphangiomyomatosis
Radiographic honeycomb lungs
Infantile spasms
Myoclonic, tonic or atonic seizures
Immediate relative with tuberous sclerosis

Patients with TS may present with purely visceral lesions: renal, cardiac or pulmonary lesions. The correct diagnosis in these cases may be difficult especially in the incomplete form of the disease with no cerebral impairment as in the case reported or with no facial eruptions. The visceral lesions may present with acute complications such as haematuria, retroperitoneal haemorrhage as in the present case or pneumothorax. In such patients it is important to make the correct diagnosis. This is particularly important from the urological aspect. Haematuria with a bizarre unilateral filling defect in the pyelogram may lead to a nephrectomy when conservative renal surgery (enucleation, partial nephrectomy) or selective arterial embolisation may be desirable as there is a good chance that the other kidney is also affected[6,7].

Visceral lesions, often asymptomatic, may be revealed by special examinations or at autopsy. The most frequent and characteristic visceral lesions affect the kidneys, heart and lungs.

Kidneys

Two types of renal lesions occur in patients with TS: angiomyolipomas and renal cysts. They may be found independently or together; they may be unilateral, bilateral, single or multiple. Crosett (1966) estimated that renal lesions occur in 50-90% of cases[8].

Golji (1961) carried out urological examination on 47 cases of TS between 3 and 59 years of age[9]. He found pyuria in 4, haematuria in 4, albuminuria in 11 and a palpable renal mass in 4. Retrograde pyelography showed normal excretory passages in 24, space occupying lesions in 9, malformations of excretory passages in 10 and chronic pyelonephritis in 2.

Renal angiomyolipomas may be totally asymptomatic and are usually clinically manifested by recurrent haematuria, flank or lumbar pain or palpable mass. They may present insidiously with progressive renal failure leading to uremia or abruptly with acute abdominal pain and hypovolemic shock after a massive retroperitoneal haemorrhage, as was the case in the present patient.

Heart

The most frequent and characteristic type of tumour is rhabdomyoma. In one series[10] of 43 children with TS, cardiac rhabdomyomata was found in 58% of subjects. Only 18% of adults had these tumours, suggesting that the tumours tend to regress in early infancy and adolescence. Cardiac symptoms

include outflow-tract obstruction, dysrhythmias and impairment of ventricular wall contractility.

Lungs

Cystic disease of the lungs and pulmonary lymphangiomyomatosis are the two possible manifestations of TS in the lungs and account for 9% of visceral manifestations. Pulmonary lesions affect women especially. Seventy-five percent of the patients are between 20 and 40 years old. In a study involving 20 cases of TS with pulmonary lesions, 15 were women and 15 were between 20 and 40 years of age. Clinical symptoms included: progressive dyspnoea in 10 cases, spontaneous pneumothorax in 7 and haemoptysis in 3[3]. Chest radiograph may reveal a honeycomb appearance.

In conclusion, it is important to recognise the acute visceral manifestations of TS, so that serious errors in diagnosis may be avoided especially in the incomplete forms of the disease without cerebral impairment or facial eruptions.

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