RADIONUCLIDE IMAGING OF THE KIDNEY IN TUBEROUS SCLEROSIS

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Two patients, related to each other, are presented who have asymptomatic tuberous sclerosis. Utilizing radionuclide imaging techniques, multiple lesions of the kidney in one and a single lesion in the other were shown to be vascular rather than cystic in nature. Because of their known high association with tuberous sclerosis, renal angiomyolipoma was the primary consideration. This was proven with angiography. It is proposed that such nuclear medicine studies may offer diagnostic information in tuberous sclerosis with little hazard or inconvenience to the patient.

Tuberous sclerosis is a dominant, inherited neurocutaneous disorder that is clinically associated with mental retardation, epileptiform seizures, and characteristic skin lesions which are adenoma sebaceum (*1*). Brain lesions, most commonly subependymal astrocytomas, have been demonstrated with both roentgenologic (*2*) and radionuclide (*3*) techniques. The disease often has multisystem involvement and tumors in other organs, e.g., heart, lung, and bone, have been encountered. Perhaps the most frequent extraneural site of involvement has been the kidney where angiomyolipoma predominates. Although this finding has had several roentgenologic descriptions (*4*), this represents the first known radionuclide demonstration of such lesions in patients with tuberous sclerosis. The clinician caring for the patient as well as the radiologist and nuclear medicine physician should be familiar with the frequent occurrence of renal angiomyolipoma in this disease and should, therefore, consider the use of radionuclide imaging studies to confirm its presence. As a result, the patient need not be subjected to more costly and hazardous studies such as renal angiography.

CASE 1

A 40-year-old white man was admitted to Fordham Hospital on October 23, 1973 for evaluation of enlarged kidneys and skin lesions over his face. The skin lesions on his face were present since early childhood. There was no history of mental retardation, seizures, or urinary tract infection. Past history indicated that in 1956 the patient sustained a minor injury at work and developed a massive retroperitoneal hematoma which was evacuated surgically. The family history disclosed that he had two children both exhibiting the same facial skin lesions. Physical examination revealed a healthy individual with erythematous papules over the nose, face, and chin. A few scattered lesions were noted on the neck, shoulders, and right foot. Neurological and psychological evaluation was entirely normal. Laboratory data indicated no significant hematological or urinary findings. X-ray examinations demonstrated cortical thickening of several metacorpals and phalanges as well as intracranial calcifications. A urogram showed bilateral enlarged kidneys with marked deformity to the collecting systems (Fig. 1). A nephrotomogram suggested possible polycystic disease involving the right kidney. Radionuclide imaging studies with ^{197}Hg-chlormerodrin and ^{99m}Tc-pertechnetate demonstrated the masses present in both kidneys to be quite vascular (Fig. 2). Selective renal angiography was performed which showed the large vascular lesions within both kidneys (Fig. 3). These appeared to be quite extensive and compressed the renal parenchyma. The appearance was typical of bilateral renal angiomyolipomas.

CASE 2

A 12-year-old boy was admitted to Fordham Hospital on October 23, 1973 for evaluation of dark pigmented skin lesions over the face that were present since 8 years of age. He is the son of the previous patient (Case 1). The patient was apparently well

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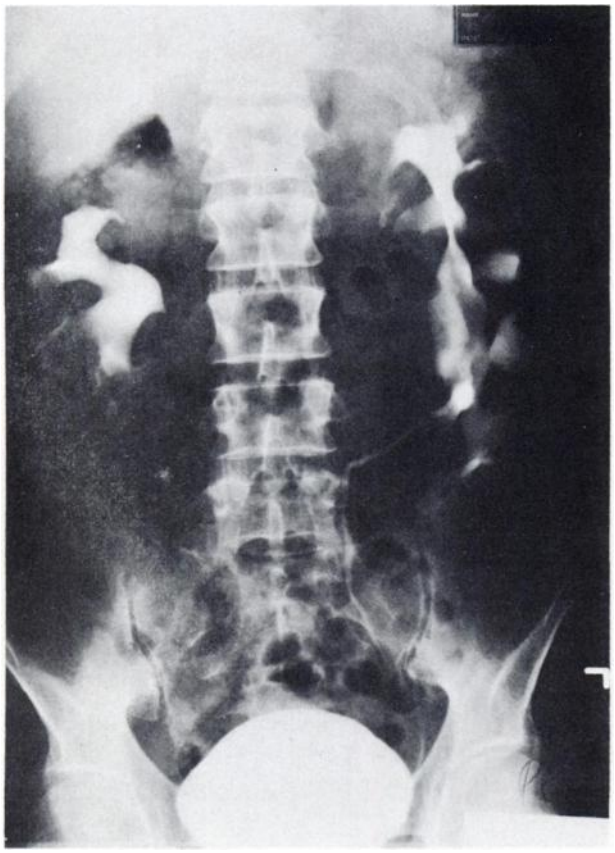


FIG. 1. Case 1. Intravenous urogram shows bilateral distortion and displacement of collecting system, somewhat more marked on left.

until about 8 years of age when he noticed red dots over his face that subsequently became larger and more deeply colored. He gave no history of seizures, genitourinary tract infection, or apparent difficulty in school. Past history was of no significance. The physical examination revealed an essentially healthy boy except for the findings of erythematous papules over the nose, molar eminences, and chin. Psychological evaluation revealed his mental ability in spelling to be consistent with that of an 8-year-old child and with that of a 9-year-old in reading. Laboratory data indicated no abnormal hematological or urinary findings. The EEG was normal. The anthropometric chart showed a child in the 40 percentile level for height and weight. X-ray examinations demonstrated a slightly enlarged calvarium; the other bony structures were normal. A urogram showed a mass in the left superior pole deforming the upper collecting system (Fig. 4). A nephrotomogram suggested that a solid lesion and not a cyst was present. Radionuclide imaging studies demonstrated the mass to be quite vascular (Fig. 5). A selective renal arteriogram was performed that showed a largely vascular lesion with numerous draining capsular veins (Fig. 6). The tumor vessels were small and aneurysmal in appearance. The appearance was typical of angiomyolipoma.

DISCUSSION

Tuberous sclerosis may occur with normal intelligence and, therefore, unless the patient has skin lesions or seizures the clinical diagnosis may long be masked. Roentgenologic examinations have played a major role in establishing the diagnosis in many of these cases.

The renal angiomyolipomas have been particularly characteristic with the reported incidence ranging from 50–80% in cases of tuberous sclerosis (*5*). They are diffuse and primarily bilateral (Case 1). However, solitary or unilateral tumors do occur (Case 2). Essentially, they are hemartomatous-type lesions with vascular and fat and, to a lesser degree, muscle components. When tumors are bilateral, the patient may be asymptomatic or may have spontaneous retroperitoneal hemorrhage. The latter finding is reported more frequently in solitary tumors (*6*). When tumors are bilateral and contain large quantities of fat or vascular components, they distort the collecting system. The mistaken diagnosis of polycystic kidney is frequently made. Interestingly, there is a dispute among investigators as to the validity of reports which indicate the coexistence of these two entities in the same kidney. Since cystic changes are described in renal angiomyolipomas, the reported

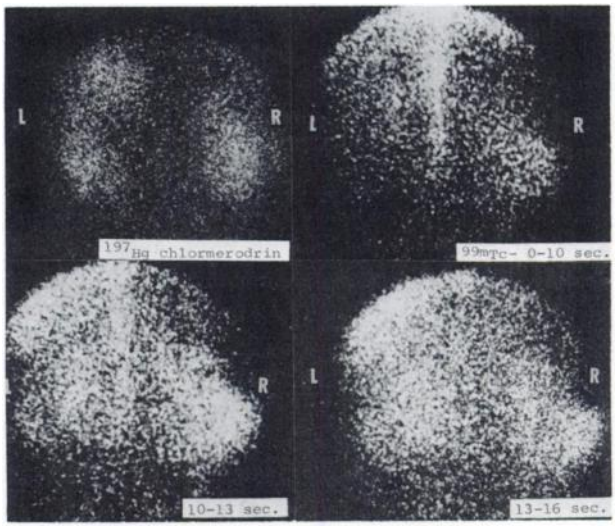
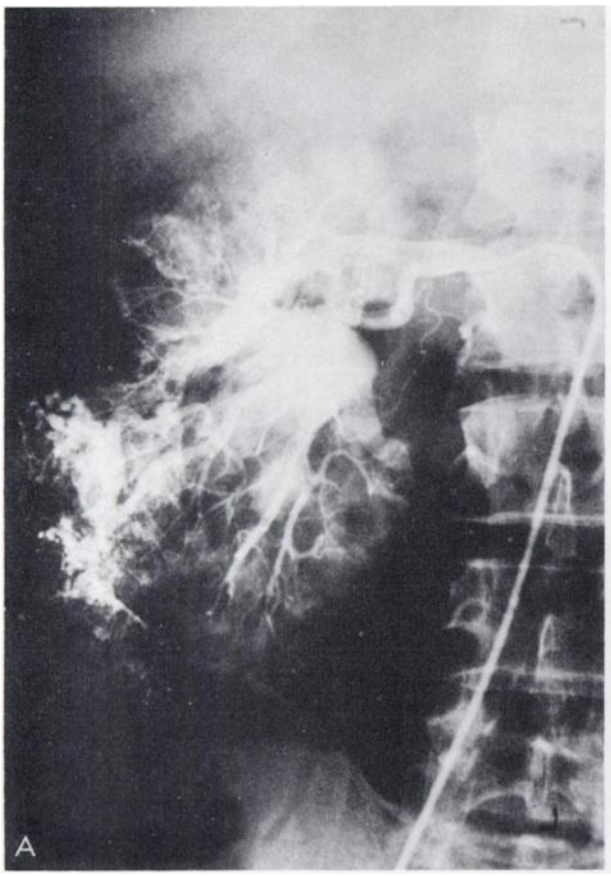


FIG. 2. Case 1. Posterior ^{197}Hg static scintiphoto shows diminished uptake in medial portion of larger left kidney as well as some diminished uptake in upper portion of right kidney. Rapid sequential scintiphotos in posterior position after bolus of ^{99m}Tc-pertechnetate show rich vascular flow to bilateral renal masses; particularly vascular lesion is noted in infralateral portion of right kidney.



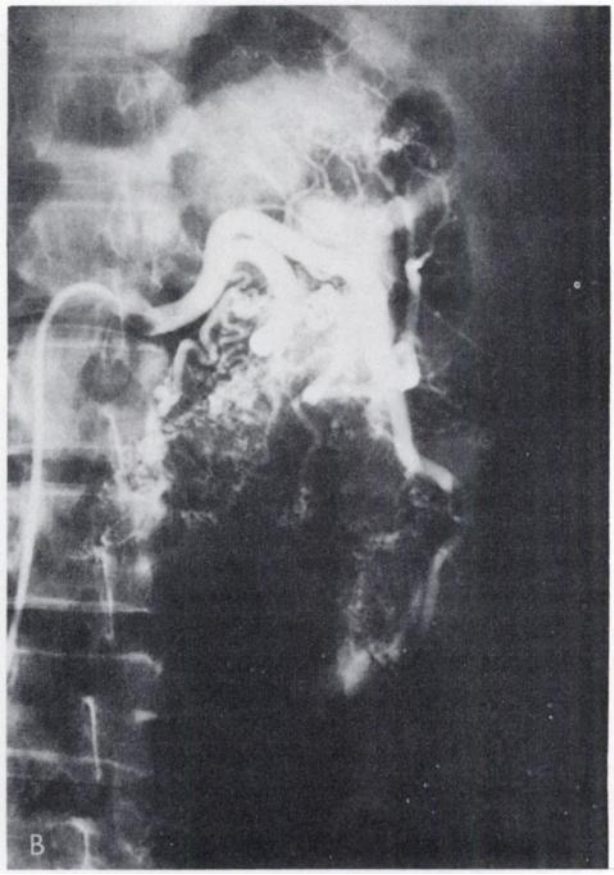


FIG. 3. (A) Case 1. Right selective renal angiogram shows markedly abnormal tumor-like vessels at infralateral portion of kidney with associated abnormal, but not as extensive, vasculature throughout remainder of kidney particularly in superolateral portion. Cystic changes are also apparent in lower portion of kidney. (B) Case 1. Left selective renal angiogram shows abnormal collection of tumor-like vessels in medial portion of kidney in region of hilus. As on right side one may note other diffuse vascular changes in remainder of kidney. Appearance is typical of angiomyolipoma.

instances of coexistent polycystic kidneys and angiomyolipomas may, in effect, be a variant of diffuse tuberous sclerosis (7).

It should be pointed out that it is extremely rare for angiomyolipomas to result in renal failure. This latter finding is common in renal polycystic disease. As in Case 1, extensive angiomyolipomas of the kidney may occur without major loss of renal function (7). The explanation for the preservation of renal function in view of extensive parenchymal damage is unknown.

The radionuclide studies require the availability of a scintillation camera. After the original demonstration of focal defects on static ^{197}Hg-chlormerodrin study, a bolus of ^{99m}Tc-pertechnetate was administered. Rapid sequential scintiphotos every few seconds clearly demonstrated activity in these lesions establishing their vascular nature (Figs. 2 and 5). Conversely, the avascular lesions of polycystic kidneys would not fill in on such "flow" studies. It is also possible to perform both of these studies with one injection of ^{99m}Tc/iron ascorbate or chelate (DTPA).

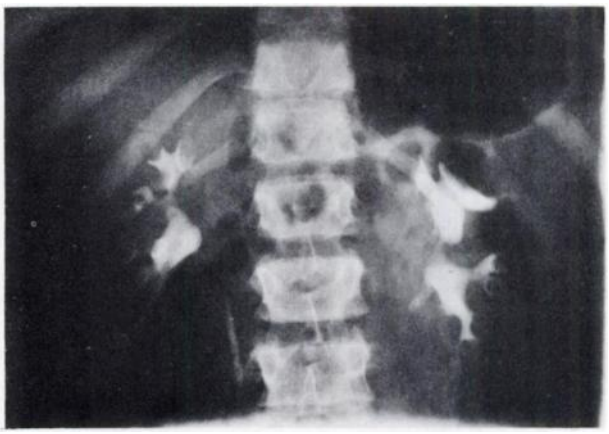


FIG. 4. Case 2. Intravenous urogram reveals flattening and slight displacement of upper pole calyx on left.

This would require performing the bolus "flow" study before the static images. This is not the optimal order of examinations since it is more desirable to first see the defects on static scintiphotos before trying to determine if they are vascular or avascular.

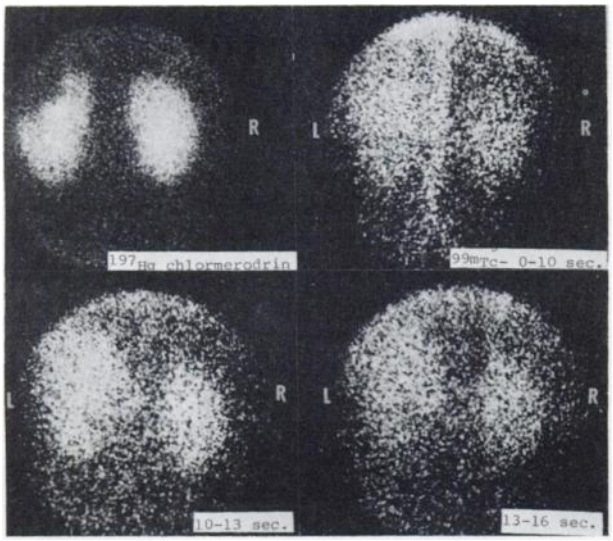


FIG. 5. Case 2. ^{197}Hg scintiphoto shows defect in upper outer portion of left kidney. Subsequent dynamic flow study after bolus of ^{99m}Tc-pertechnetate shows that lesion is distinctly vascular. Abundant activity fills abnormal area.

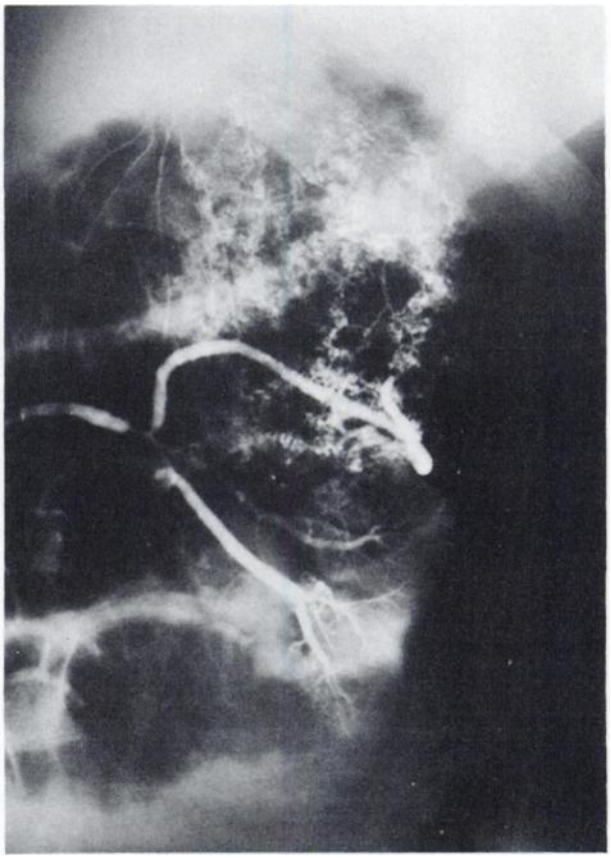


FIG. 6. Case 2. Left selective renal angiogram shows extensively vascular mass at upper pole of left kidney, which is quite typical of angiomyolipoma. Incidentally noted is presence of vascular spasm at tip of catheter in renal artery.

The value of the radionuclide studies lies in their ability to demonstrate that single or multiple vascular lesions of the kidney are present in a patient with tuberous sclerosis. Particularly if multiple in nature, the diagnosis of angiomyolipoma can then be established with a reasonable degree of certainty. Conventional radiographic angiography would not be necessary. Since children are often involved, this is quite important. Although performed in these two cases to confirm the radionuclide findings, we would not use angiography in similar situations in the future.

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