Case Report

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Ultrasound and Color Flow Doppler Manifestations of Pseudoxanthoma Elasticum

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Pseudoxanthoma elasticum (PE) is an inherited disorder of elastic tissues characterized by progressive protean clinical manifestations. The most frequently encountered manifestations include xanthoma-like papular and reticulated skin lesions, retinal angioid streaks and other chorioretinal changes that often impair vision, hemorrhage from the upper gastrointestinal tract, occlusive peripheral and coronary vascular disease, hypertension, uterine bleeding, and mucosal lesions.[1–3]

CASE REPORT

A 31-year-old man was hospitalized to determine the cause of upper gastrointestinal hemorrhage. He had had four episodes of gastrointestinal hemorrhage within the past 3 years. He had no abnormal bleeding tendency and the coagulation screen was negative. Gastroscopic examination showed multiple erosions around the cardia and on the anterior wall of the corpus.

A decrease in the vision of the right eye had been noted 6 months prior to his present admission. Examination of the fundi revealed bilateral angioid streaks and an atrophy of the upper half of the right optic disc.

Upper abdominal ultrasonography demonstrated diffuse multiple hyperechoic foci, measuring 2 mm to 3 mm in diameter, within the renal parenchyma. The spleen had a similar but more sparsely distributed dotted echographic pattern (Figures 1A and 1B).

Color flow ultrasonography showed both morphologically and hemodynamically normal renal arteries. In fact, the arterial blood pressure of the patient was regular. Pulsatility and resistive indices obtained from the interlobar and arcuate arteries were within normal limits (Figure 2).

Peripheral color Doppler ultrasonography was performed because of his complaint about muscular pain of the lower extremities produced by walking and relieved by rest. This study found a complete occlusion of right superficial femoral artery starting from its proximal segment (Figure 3). In the distal part of the left superficial femoral artery, spectral Doppler examination showed doubling of the peak systolic velocity indicating 50% to 99% stenosis. No color flow nor image-directed Doppler signals were obtained from the left popliteal artery (Figure 4). A peripheral arteriogram confirmed the findings of the color Doppler ultrasonography.

Plain films of the abdomen and thorax, proctoscopic examination, and blood and urine analysis were normal.

DISCUSSION

Pseudoxanthoma elasticum occurs with an incidence of about one in 160,000 individuals.[4] The pattern of inheritence is generally assumed to be autosomal recessive although autosomal dominant forms have been identified.[3–5]

Histopathological studies have shown that calcification of the elastic fibers represents the earliest abnormality in PE. Arterial calcifications are common in this systemic disease, and the media is the predominant layer of calcium deposition. The patients are prone to vascular degenerative processes, primarily observed in peripheral, renal, and coronary arteries. Intermittent claudication and renovascular hypertension are well-known complications of the disease.[1]

In our case, we examined the arteries of the lower extremities with color flow Doppler imag-

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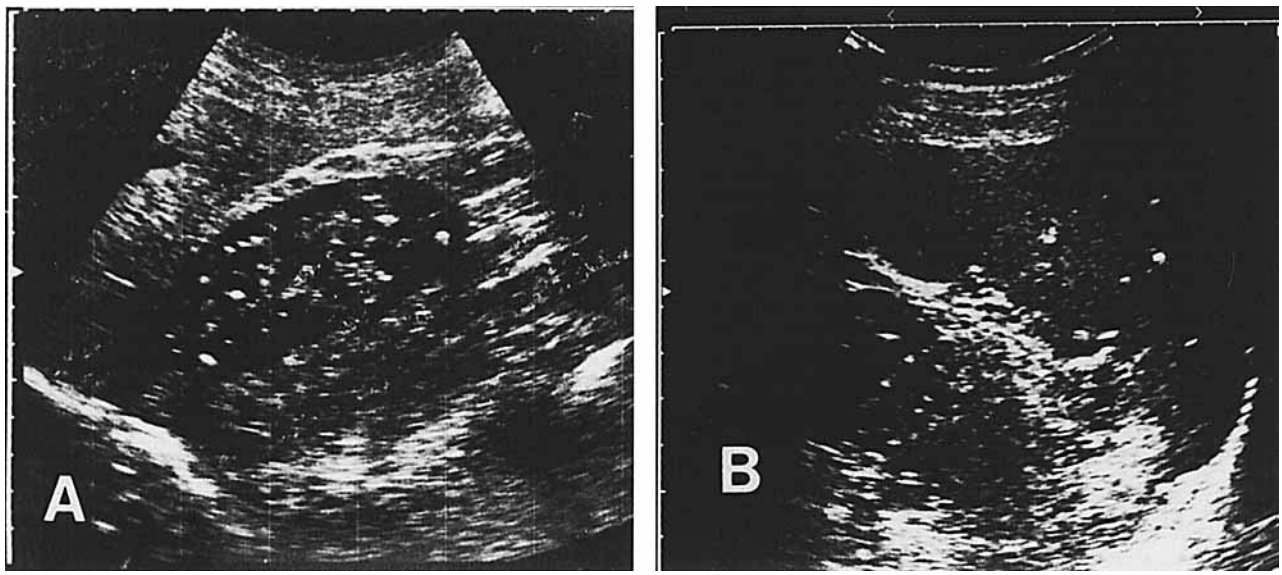

FIGURE 1. Dotted hyperechogenic pattern in the right kidney **(A)** and spleen **(B)**.

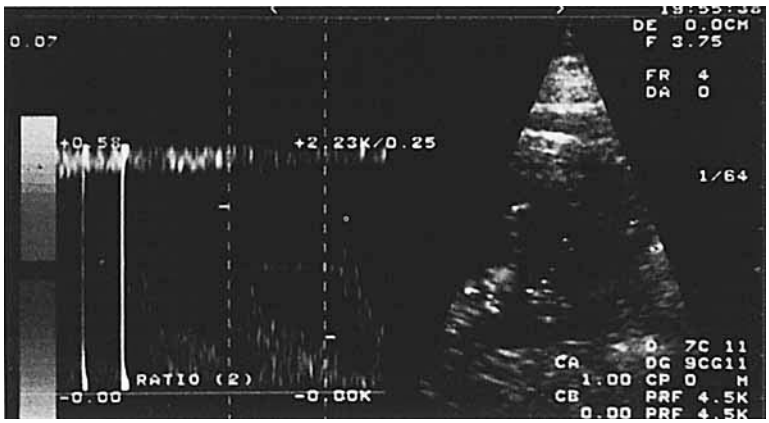

FIGURE 2. Color flow imaging and Doppler spectrum obtained from the interlobar and arcuate arteries were normal.

ing to demonstrate the localization, degree, and extension of the peripheral arterial disease. Occlusive and stenotic lesions were detected in both legs, compatible with the patient's claudication-like complaints. An important point related to the patients with PE is low elasticity of their tissues that are most likely to disrupt during stretching. Thus, various therapeutic procedures, e.g., transluminal balloon angioplasty, may have an increased risk of vessel disruption compared with other patients without PE.[4]

The underlying elastic tissue defect produces markedly altered physical properties in many organ systems. Gastrointestinal hemorrhage usually occurs early in the course, at a time when other clinical manifestations are minimal. Development of retinal angioid streaks and subsequent chorioretinal changes may severely diminish visual ability. The xanthoma-like papular skin lesions on the flexural surfaces of the extremities and redundant folds of lax and flabby skin, particularly about the neck and axillae, are the most striking defects of this disorder.[1–3] In

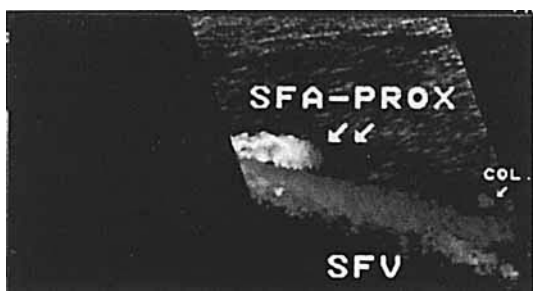

FIGURE 3. Occlusion of right superficial femoral artery.

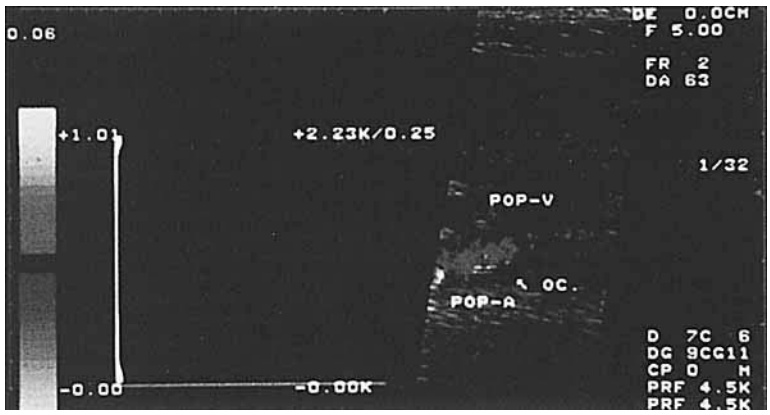

FIGURE 4. Absence of color-flow and image-directed Doppler signals in the left popliteal artery.

our case, no skin changes compatible with PE were present.

A characteristic sonographic pattern of dotted increased echogenities of the interlobar and/or arcuate arteries in both kidneys has been shown by Gubler et al. in two children with PE. Renal biopsy disclosed arterial calcification located within the media.[6] In our case, this dotted sonographic appearance of the kidneys played an important role in the diagnosis of PE. However, the patient refused renal biopsy. Similar, but more sparsely distributed, hyperechogenic spots were seen in the spleen. Suarez et al. demonstrated a similar dotted appearance in the spleen and pancreas as well as in the kidneys in a normotensive pediatric patient. These investigators believed these hyperechogenities represented a calcifying process involving the elastic layers of the arteries in these viscera.[7] Reflecting the characteristic of the disease, calcium deposition in the walls of the abdominal arterial branches is known to occur in patients with PE.[1]

Intraparenchymal calcifications corresponding to the sonographic findings cannot be detected in plain abdominal films. Although computerized tomography (CT) was not performed on our patient, in the pediatric case of PE reported by Suarez et al., it is stated that CT has been unable to detect these parenchymal calcifications.[7]

We used color flow imaging as a noninvasive and new method to evaluate the renal arteries and perfusion of both kidneys. Because renovascular disease was not so severe in our patient, both renal arteries were found to be morphologically and hemodynamically normal. The Doppler spectrum obtained from interlobar and arcuate arteries were within normal limits despite the calcifying process in these vessels, indicating that vascular abnormalities had not progressed to the point where alterations in renal blood flow had occurred.

Our findings lead us to conclude that detecting multiple, diffuse, small, hyperechogenic foci within the renal parenchyma is a very significant sonographic sign of PE. As it is in our case, having had this rare but typical pattern clarified, the results of other examinations (fundoscopy, gastroscopy, peripheral Doppler imaging) subsequently assisted in the accurate diagnosis of PE.

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