

Cystic Changes of Breast in a Family With Autosomal Dominant Polycystic Kidney Disease

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Autosomal dominant polycystic kidney disease is associated with cysts in many organs including the liver, pancreas, lungs, spleen, ovaries, testes, thyroid, and uterus. However, there is no report, to our knowledge, of cystic changes of the breast along with this disease. We describe 3 members of a family with multiple bilateral breast cysts in association with autosomal dominant polycystic kidney disease.

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INTRODUCTION

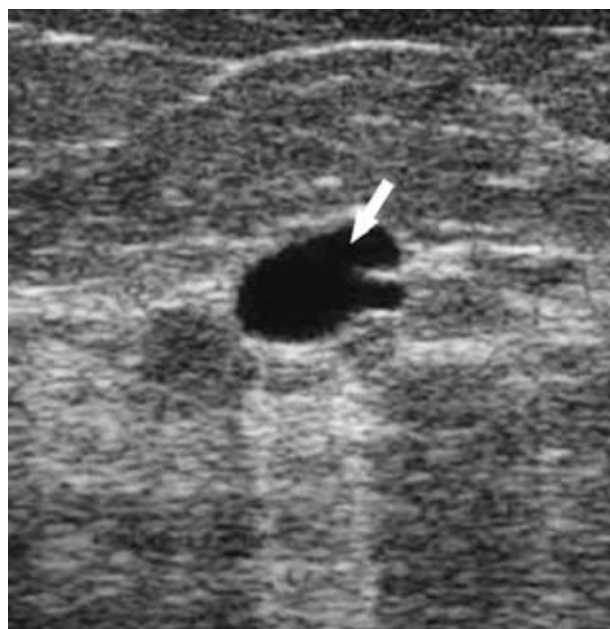
Autosomal dominant polycystic kidney disease (ADPKD) is a common disorder, occurring in approximately 1 in every 400 to 2000 live births.^{1,2} It is associated with cysts in many organs including the liver, pancreas, lungs, spleen, ovaries, testes, thyroid, uterus, and bladder.²⁻⁵ However, to our best knowledge, the occurrence of breast cysts in patients with ADPKD has not been reported previously. Herein, we present 3 members of a family with multiple bilateral breast cysts and ADPKD.

CASE REPORT

A 21-year-old college student girl presented to our center for evaluation of bilateral breast masses. General physical examination of the patient was unremarkable. On breast examination, there were no dysmorphic or abnormal cutaneous findings, but the patient had a 30 × 30 × 50-mm mass in her right breast and a 20 × 20 × 20-mm mass in her left breast. Concerning family history, her paternal grandfather and her brother had died due to kidney failure. Her 59-year-old father and 2 sisters had undergone kidney transplantation because of end-stage renal disease due to ADPKD. One of her sisters was evaluated for bilateral breast masses 2 years after kidney transplantation, and multiple bilateral breast cysts were confirmed by mammography and ultrasonography (Figure). She refused aspiration of the cyst and accepted to be followed up. She

had no problem to date except for transient pain in her breast, responding to acetaminophen. Another kidney transplanted sister of the patient had also the same bilateral breast cysts.

Regarding the family history of ADPKD, abdominal and breast ultrasonography was obtained for possible association of breast cysts and ADPKD in the nontransplanted patient, which showed renal cortical cysts, a small liver cyst, and multiple



A simple breast cyst in the patient with autosomal dominant polycystic kidney disease.

bilateral breast cysts leading to the diagnosis of asymptomatic ADPKD. Aspiration of breast cysts was considered, but she refused it and only accepted to be followed up. In her 6th month of follow-up, the cysts had persisted, inducing some pain in her right breast that contained the largest cyst.

DISCUSSION

Cysts are a common cause of breast lumps in premenopausal women 40 to 49 years of age; 63% of breast cysts are detected among these women.⁶ About 20% to 25% of palpable breast abnormalities are simple cysts.⁷ The differential diagnosis of a dominant breast abnormality commonly includes cysts, fibroadenoma, benign nonproliferative lesions, benign proliferative lesions with or without atypia, fat necrosis, ductal carcinoma in situ, and invasive cancer. The initial objective is to distinguish simple cysts from solid masses, because simple cysts are benign and do not require further evaluation.^{6,8,9}

Breast cysts are reported in kidney transplant recipients and considered to be due to cyclosporine A in these patients.^{10,11} However, one of our patients had functioning native kidneys and did not have a history of medication use, including cyclosporine A. Therefore, cyclosporine A could not be considered as a cause.

In one study, the contributions of amino acids to the overall osmotic activity of cyst fluids were sought in fluids from the renal cysts of patients with ADPKD and gross cystic disease of the breast. Fluids from 18 women with gross cystic disease of the breast and from 8 patients with ADPKD, grouped on the basis of sodium concentrations, were analyzed for potassium, chloride, protein, and osmolality by routine methods and for amino acids by reversed-phase high-pressure liquid chromatography. There were similarities between kidney and breast cyst fluids, including relative isosmolality, a wide range of sodium concentrations, an inverse relationship between sodium and potassium concentrations, higher concentrations of amino acids in low-sodium fluids, and significant correlations between sequential concentrations of amino acids in cyst fluids versus blood and some of its components. Therefore, cyst fluids in ADPKD and gross cystic disease of the breast share compositional characteristics.¹²

In another study, the association between

polythelia (supernumerary nipple) and kidney and urinary tract malformations was assessed.¹³ The study was performed in 146 white patients (123 men and 23 women) with accessory mammary tissue out of 2645 participants consecutively referred for physical examination. Kidney and urinary tract malformations were detected in 11 patients with accessory mammary tissue (9 men and 2 women) and in 1 participant in the control group. These indicated a significantly higher frequency of kidney and urinary tract malformations in patients affected by accessory mammary tissue compared to controls (7.5% versus 0.7%, $P < .001$). Also, a broad spectrum of kidney and urinary tract malformations was discovered in association with accessory mammary tissue including ADPKD, unilateral renal agenesis, cystic renal dysplasia, familial renal cysts, and congenital stenosis of the ureteropelvic junction. The authors concluded that accessory mammary tissue offered an important clue for congenital and hereditary anomalies of the kidneys and urinary collecting systems. They suggested that patients with accessory mammary tissue be extensively examined for the presence of occult nephro-uropathies.¹³

Based on these findings, we suggest that cystic changes of the breast in this family were associated with ADPKD. To our knowledge, this is the first report of familial occurrence of multiple bilateral breast cysts in association with ADPKD. Further studies are necessary to clarify this association.

CONFLICT OF INTEREST

None declared.

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