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Case Report

Incidental extra cystic findings in autosomal dominant polycystic kidney disease: beyond the liver and kidney

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ABSTRACT

Autosomal dominant polycystic kidney disease (ADPKD) is a genetic disorder primarily affecting the kidneys and liver, but its systemic nature can lead to unexpected extrarenal findings. This report describes a 55-year-old woman with ADPKD who presented with hypertension and abdominal discomfort. Diagnostic imaging uncovered not only typical renal and hepatic cysts but also rare incidental lesions-a caecal lipoma, ovarian dermoid, and diverticulum. Surprisingly, she later developed synchronous bilateral breast cancer, suggesting a possible link between ADPKD and malignancy. These findings highlight the need for comprehensive evaluation in ADPKD patients, as extrarenal manifestations may signal underlying comorbidities. A multidisciplinary approach is essential for early detection and management of such complex presentations, emphasizing the importance of regular surveillance beyond renal and hepatic involvement.

Keywords: Autosomal dominant polycystic kidney disease, Caecal lipoma, Extrarenal manifestations, Incidentaloma, Ovarian dermoid, Synchronous breast cancer

INTRODUCTION

Autosomal dominant polycystic kidney disease (ADPKD) is caused by PKD1/PKD2 mutations, leading to progressive renal cysts and end stage renal disease. While renal/liver involvement is well-documented, extrarenal manifestations-including cardiovascular defects, diverticula, and benign incidentalomas-are underreported.[1] Incidental findings (gastrointestinal lipomas, ovarian dermoid) may complicate diagnosis or signal comorbidities.[2] Rarely, ADPKD coexists with malignancies, though causality remains debated.[3] We present a case highlighting ADPKD's multi-system complexity, featuring incidental benign lesions and synchronous breast cancer.

CASE REPORT

A 55-year-old female presented with complaints of progressive abdominal distension and dull, aching lower back pain for 6 days, accompanied by reduced appetite and new-onset vomiting for 1 day. There were no fever, hematuria, or urinary symptoms. Past History was significant with hypertension diagnosed 3 years prior however patient was on irregular treatment. No known renal disease or family history of ADPKD and no prior surgeries. On clinical examination, patient was afebrile, blood pressure was 180/100 mmHg and pulse rate 88 rate beats per minute. Per abdomen examination showed diffuse tenderness, no palpable masses or organomegaly. Cardiovascular examination showed S4 gallop; no murmurs. Other systems were Unremarkable. Complete hemogram with peripheral smear showed hemoglobin of 10.2 g/dl (normocytic normochromic), renal function: urea 23 mg/dl, creatinine 2.32 mg/d, urinalysis: albumin 1+, no active sediment, electrolytes revealed sodium 135 Meq/l, potassium 4.8 Meq/l. Ultrasound abdomen/pelvis revealed bilaterally enlarged kidneys with multiple anechoic cysts (largest 48 mm in left lower pole), liver showed dilated intrahepatic biliary radicals in left lobe, multiple hepatic cysts (largest 13 mm in segment IV a). Contrast-enhanced CT (CECT) abdomen/pelvis showed innumerable renal

bilateral cysts (3–48 mm), no solid enhancing lesions, multiple hepatic cysts; left intrahepatic duct dilatation. Colonoscopy revealed fat-attenuating submucosal lesion in the cecum (22 mm), well-circumscribed left adnexal mass (3.0×2.7 cm) fat-fluid level, air-filled outpouching in sigmoid colon suggestive of diverticulum (Figure 1).

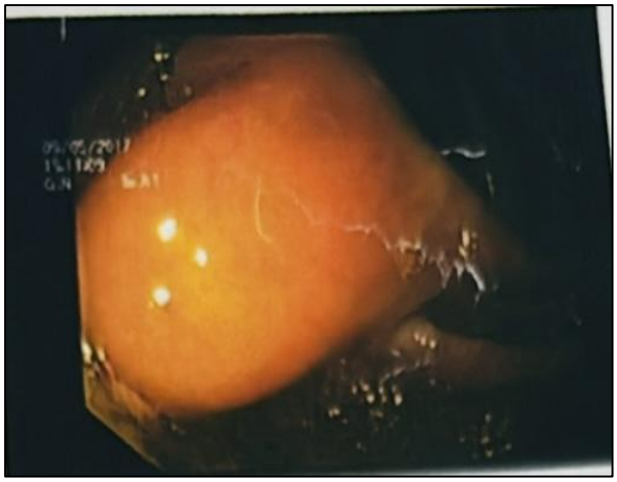

Figure 1: Colonoscopy showing Caecal submucosal lipoma measuring 22 mm.

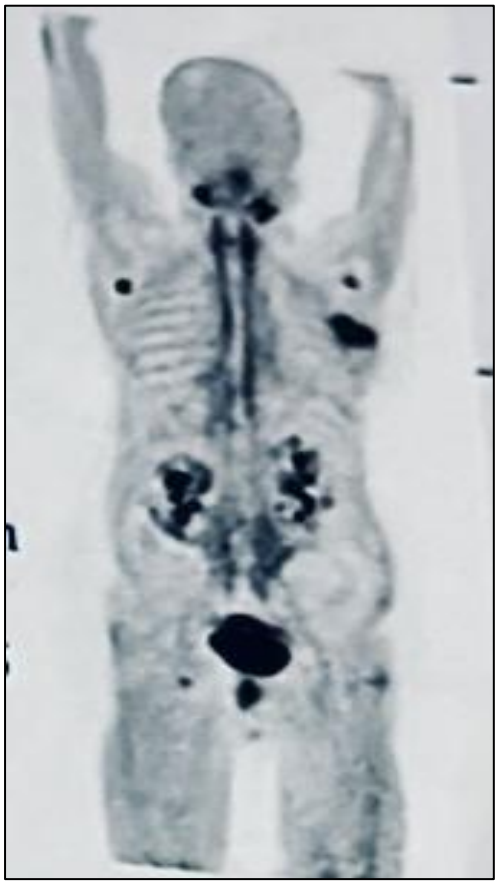

Figure 2: PET-CT showing bilateral enlarged kidneys showing polycystic morphology, hepatic cysts, few enlarged axillary, pectoral nodes and soft tissue lesion in both breasts and ovary.

Echocardiography revealed sclerotic aortic valve, left ventricular hypertrophy, grade I diastolic dysfunction. Hypertension was managed with Intravenous labetalol transitioned to oral amlodipine. Blood pressure was stabilized to 130/80 mmHg. Patient was discharged on day 7 with planned haemodialysis initiation (eGFR 18 ml/min/1.73m²). Patient was followed-up 2 months later presented with new onset firm bilateral breast lumps (left> right). On examination, left breast mass (4 cm, upper inner quadrant).

Fine-needle aspiration (FNAC) was suspicious for malignancy (Bethesda V). Tru-cut Biopsy showed Invasive ductal carcinoma (ER+, PR+, HER2-). Right breast mass (1.2 cm, upper quadrant). Core biopsy revealed Synchronous invasive ductal carcinoma. PET-CT revealed Left breast 3 cm FDG-avid mass with skin thickening/extension and Right breast of 1.2 cm FDG-avid nodule. Bilateral axillary lymphadenopathy largest measuring 1.5 cm and ovarian dermoid as described in Figure 2.

Patient was managed with neoadjuvant chemotherapy (4 cycles adriamycin cyclophosphamide followed by 12 cycles of paclitaxel). Later modified radical mastectomy (left) and lumpectomy (right) with adjuvant radiotherapy was done. Patient was stable on further follow-ups.

DISCUSSION

This case underscores that ADPKD manifests beyond renal and hepatic cysts, with significant extrarenal findings including benign incidentalomas (caecal lipoma, ovarian dermoid,) and a synchronous malignancy. While gastrointestinal lipomas and ovarian dermoid are typically incidental, their co-occurrence with ADPKD suggests potential shared pathways in cellular proliferation or extracellular matrix defects.[4]

The rapid emergence of bilateral breast cancer shortly after ADPKD diagnosis highlights a diagnostic challenge. Multi-disciplinary approach is needed to address complex presentations especially when incidental findings or new symptoms arise during the routine ADPKD surveillance.

CONCLUSION

ADPKD requires systemic evaluation extending beyond cyst monitoring. Incidental extrarenal lesions (e.g., GI lipomas, adnexal masses) require surveillance and routine screening. This case advocates for a holistic imaging review protocol in ADPKD and reinforces early cross-specialty collaboration to optimize outcomes in this multisystem disorder, cancer screening in ADPKD with atypical presentations.

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