Indian Journal of Pathology and Oncology 2023;10(4):417–419

Content available at: https://www.ipinnovative.com/open-access-journals

Indian Journal of Pathology and Oncology

Journal homepage: www.ijpo.co.in

Case Report

Autosomal dominant polycystic kidney disease coming up with an unusual presentation of renal cell carcinoma on its first encounter

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ARTICLE INFO

Article history:
Received 08-11-2023
Accepted 25-11-2023
Available online 11-12-2023

Keywords:
ADPKD
Renal cell carcinoma

ABSTRACT

Autosomal polycystic kidney disease (ADPKD) is a hereditary condition characterized by expanding cysts that destroys the renal parenchyma causing ultimately the renal failure. The role of ADPKD in causing Renal cell carcinoma (RCC) is still under discussion. A 40-year-old male came with fever with chills with history of hematuria, flank pain and weight loss for 2 months. A CT urography showed Bilateral adult polycystic kidney disease and Upper pole of right kidney showing large well-defined lobulated heterogeneously enhancing mass measuring 11x10.5x11.4 cm, most likely renal cell carcinoma. Patient underwent open right radical nephrectomy. On histopathology, diagnosis was given as Papillary Renal cell carcinoma, background kidney showing features of Autosomal polycystic kidney disease. This case report comprehensively described the coexistence of Adult polycystic kidney disease with renal cell carcinoma, which is unique presentation to present with.

1. Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is one of the most common hereditary kidney disorders occurring in about 1 or 2 per 1000 live births. The pattern of heritance shows complete penetrance and each offspring of an individual has 50% chance of inheriting the disease. It is caused by mutation in PKD 1 and PKD2 gene mutation implicated in regulation of cell cycle and ciliary function.

Renal cell carcinoma (RCC) is a kidney malignancy that accounts for 5% of male and 3% of female cancer cases globally.[1] According to World Health Organization, there are more than 1,40, 000 RCC-related deaths yearly and RCC ranks as the 13th most common cause of cancer death.

The link between ADPKD and RCC has been postulated, but due to limited number of cases, currently ADPKD is not held as a risk factor for the development of RCC.[2]

This case report adds one more incident of coexistence of ADPKD and RCC.

2. Case Presentation

A 40-year-old male came with fever with chills with history of haematuria, flank pain and weight loss for 2 months. Patient was a chronic tobacco chewer and smoker. There was no family history of malignancy or cystic disease. A CT urography showed a large well defined lobulated heterogeneously enhancing mass measuring 11x10.5x11.4 cm arising from the upper pole of right kidney, with areas of necrosis and cystic changes seen within. Multiple enlarged heterogeneously enhancing precaval, preaortic, aortocaval and retrocrural lymph nodes were seen, largest measuring 6.6x5.1X1.5cm in size. Both kidneys were enlarged in size with multiple cortical cysts of varying sizes. Liver showed tiny cysts in both lobes. Visualised lung parenchyma

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https://doi.org/10.18231/j.ijpo.2023.095

revealed small cysts in both lower lobes. The features were suggestive of neoplastic aetiology, most likely renal cell carcinoma of right kidney with ADPKD. Patient underwent open right radical nephrectomy. On gross examination (Figure 1), kidney was 15x11x10cm, externally bosselated. Cut section showed a solid, fleshy, grey white, tumour with variegated appearance, areas of haemorrhage and necrosis measuring 11x8x1.5 cm, predominantly in upper and middle pole. The tumour involved pelvicalyceal system. Adjacent kidney showed variable sized cysts ranging from 0.5 to 4 cm, containing serous fluid. A paracaval lymph node showing a grey white friable tumour was also received. On histopathology, a tumour arranged in papillary and solid pattern was seen with abundant eosinophilic cytoplasm and areas of necrosis. Background kidney showed multiple cysts of varying sizes lined by cuboidal to flattened epithelium, at places showing hyperplasia, and intermittent stroma showing chronic inflammatory cell infiltrate, features of Autosomal dominant polycystic kidney disease. Paracaval lymph node showed high tumour deposits. Final diagnosis was given as Papillary Renal cell carcinoma ISUP Grade III.

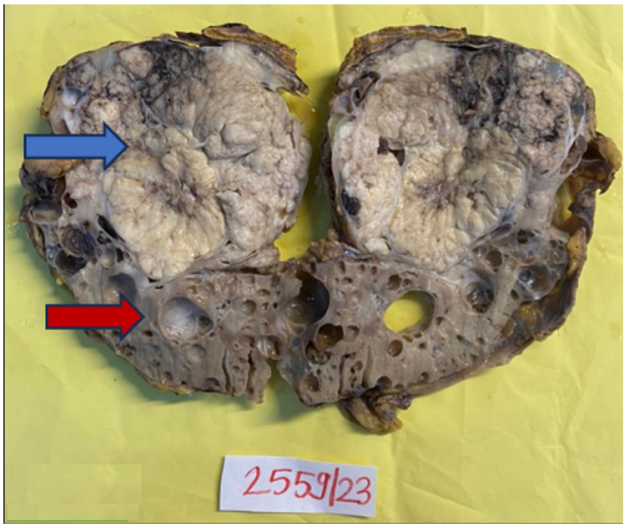

Figure 1: Blue arrow shows a tumour with variegated appearance, areas of haemorrhage and necrosis in the upper and middle part of kidney. Red arrow shows multiple cysts of varying sizes in adjacent kidney

3. Discussion

ADPKD is a hereditary condition which is characterised by progressively expanding cysts that ultimately destroys the renal parenchyma leading to renal failure. The diagnosis can be made by presence of 2 unilateral or bilateral cysts before the age of 30 years, at least 2 cysts on each kidney between the ages of 30 and 59 years, or 4 cysts on each kidney at age 60 years or older.[1] The kidneys are markedly enlarged and have bosselated outer cortical surface due to varying

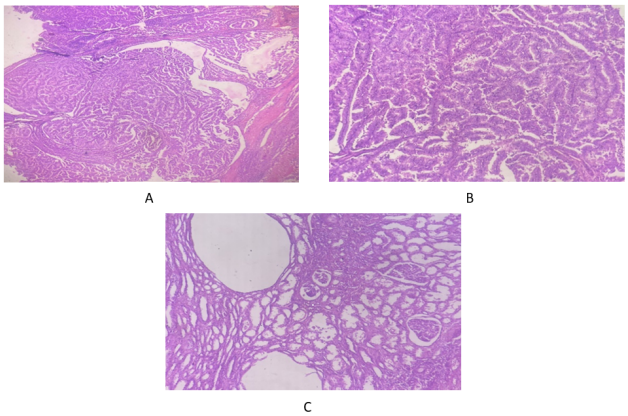

Figure 2: A): Shows papillary architecture of tumour cells; **B):** Shows high power magnification of tumour cells with eosinophilic cytoplasm accompanied by diffuse papillary pattern; **C):** Shows cysts lined by flattened epithelial cells

sized cysts. The cysts may develop in other organs such as lung, liver, spleen, or pancreas. The other manifestations include flank pain, haematuria, hypertension, flank masses and renal failure.

In this study, ADPKD was diagnosed based on the presence of atleast 2 cysts on each kidney between the age of 30 to 59 years along with cyst in liver and lungs.Also, the gross findings of involvement of cortical surface of the kidney by cysts aids in the diagnosis.

RCC is generally seen in age group of 55-60 years, with male to female ratio of 2:1. Cigarette smoking and high blood pressure are associated with increased risk of RCC. In this case, patient was cigarette smoker and tobacco chewer. A case study done by Chase C. Hansen et.al out of 16 cases of nephrectomy specimen with ADPKD, 2 cases showed presence of RCC, the median age of presentation was 54 years. The histologic subtypes identified were clear cell type and mixed (clear cell, papillary and sarcomatoid type).[2] In a case study done by Matevey T. et al. on 845 patients, current and former smokers had 1.5- and 1.6-fold increased odds of advanced RCC respectively.[3] According to the National Cancer Institute SEER program, the incidence of sporadic RCC is estimated to be 21.02 per 100,000 in men and 10.4 per 100,000 in women.[4]

A possible association between RCC and ADPKD was first suspected and described by Walters and Brasch in 1934.

According to study done by Hajj P. et al. the prevalence of RCC was higher in patients with ADPKD and End stage renal disease (ESRD), with >1 year on dialysis or renal transplantation undergoing nephrectomy according the protocol than the patients with ESRD alone (11 out of 89 nephrectomy specimens with ADPKD were showing features of carcinoma). On histology, 58.3% (7 out of 12) were showing clear cell carcinomas and 41.7% were showing tubulopapillary carcinomas. Three kidneys were

showing multifocal tumours and one patient showing bilateral tubulopapillary carcinomas.[5]

One of the hypothesis postulated for the development of RCC in ADPKD includes chronic renal injury favouring the renal parenchymal genetic mutations with consequent malignant change, or hyperproliferation in ADPKD acting as a precursor to RCC.[6] According recent WHO, Papillary renal cell carcinoma (PRCC) is more common in chronic renal disease and acquired cystic disease. Papillary Renal cell carcinoma accounts for 13-20% of renal epithelial tumours.[7]

Surgery is the mainstay of treatment in these cases along with chemotherapy or immunotherapy in cases with metastatic disease. M-TOR inhibitors with VEGF (Vascular Endothelial Growth Factor) targeted medications have also been found to be effective. The patient in this case was managed by modified radical nephrectomy. As the patient presented with RCC along with lymph node metastasis, unfortunately patient died after 2 months of treatment.

4. Conclusion

ADPKD with background of RCC is a rare presentation and poses a difficult challenge for clinician to diagnose. ADPKD patients presenting with RCC features is a rare situation but with regards to poor outcome of these cases, clinician should keep a suspicion of development of RCC along with ADPKD.

5. Source of Funding

None.

6. Conflict of Interest

None.

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Cite this article: Padmanabhan A, Masumdar AS, Gadgil N, Padalkar G. Autosomal dominant polycystic kidney disease coming up with an unusual presentation of renal cell carcinoma on its first encounter. *Indian J Pathol Oncol* 2023;10(4):417-419.