Malignant Transformation in Autosomal Dominant Polycystic Kidney Disease

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Abstract:

Background: Autosomal dominant polycystic kidney disease is one of the most common hereditary kidney disorders in adults that may develop malignant transformation. Early diagnosis of such complication is important to get rid of primary malignancy & control distant metastasis as early as possible. Sudden onset of fulminant hematuria, pain & fever in patients with end stage renal disease &/or on dialysis may raise the suspicious of such dangerous complication. This guides us to diagnose malignant renal mass by 2 D ultrasound, CT scan & histopathological evaluation. **Subject & method:** Bedside 2D ultrasound role in characterization of malignant renal mass in adult male patient in collaboration with CT scan of the abdomen & histopathological study of the specimen. **Results:** ultrasound & CT scan of the abdomen had a crucial role in characterization of malignant renal mass in patient with autosomal dominant polycystic kidney disease & end stage renal disease. **Conclusion:** Sudden onset of gross hematuria in a case of end stage renal disease must rise the concern of malignant transformation of cystic lesion in the kidney.

Keywords: Malignant, Autosomal, Dominant, Polycystic, Kidney.

Introduction:

Autosomal dominant polycystic kidney disease (ADPKD) is one of the most common hereditary kidney disorders & it accounts for 8% and 10% of patients with ESRD in the United States & Europe respectively. The mean age at diagnosis of RCC is 47 y with more predilections to males. Positive family history for ADPKD or renal failure were reported. Regarding the histologic subtype of the tumor, they include clear cell type, sarcomatoid type &

synchronous sarcomatoid & clear cell type carcinoma. (1)

In some patients with ADPKD, the kidney continues to be enlarged after the initiation of dialysis therapy, resulting in such significant complications as cyst bleeding & abdominal distension. (2)

Subject & method:

This is a case report study in a 43 y old Egyptian male patient with end stage renal disease on hemodialysis for 4 years. He was admitted for evaluation of gross hematuria, acute right flank pain & fever. This study was conducted in Radiology Department of Mansoura Health Insurance Hospital, Mansoura, Egypt, at 10-20 May, 2020 time range. Written informed patient consent for publication was obtained. 35& to 50% of patients with ADPKD had hematuria with a precipitating event such as a urinary tract infection or strenuous activity. Gross hematuria is more likely among individuals with larger kidneys (particularly when >15 cm in length), hypertension, and higher plasma creatinine concentrations. (3) There was a positive family history of ESRD & hemodialysis. On clinical examination, he was feverish, hypertensive, had abdominal distension& edema of both lower limbs. Sr. Cr. Was (7.5 gm/dl, normal range is 0.8-1.4 gm/dl), TLC is (8.5 X 1000 gm/dl, normal range is (4.5-10 X 1000 gm/dl) & otherwise unremarkable laboratory results.

Results:

Ultrasound is an excellent choice for first time diagnosis & follow-up. It's able both to suggest malignant transformation of the cystic lesion in an autosomal dominant polycystic kidney disease in adults. Abdominal ultrasound revealed marked enlargement of both kidneys, measured (22.5 X 12.5 cm) with multiple variable sized renal cysts (Fig. 1). On CCDI, the suspicious mass showed abnormal internal vascularity (Fig. 2). Right kidney showed a solitary soft tissue mass in the upper pole, measured (8.5 X 7.5 cm) with irregular borders & heterogeneous echopattern (Fig. 3). NCCT abdomen revealed cystic lesion with a mural soft tissue mass in the upper pole, measured (8.5 cm) in diameter. It's associated with infiltration of right supra-renal gland & enlarged para-aortic lymph nodes. No crossing of midline & no invasion of renal vein (Fig. 4). Imaging of patients with autosomal dominant polycystic kidney disease can be difficult due to the size and number of the cysts and associated mass effect on adjacent structures, so it is important to assess all cysts for atypical features, that may reflect complications (e.g. hemorrhage or infection) or malignancy (i.e.

renal cell carcinoma). (4) Simple renal cysts will appear anechoic with well-defined imperceptible walls, posterior acoustic enhancement. Cysts with hemorrhage or infection will demonstrate echogenic material within the cyst, without internal blood flow. Calcification may develop. Renal cell carcinomas in contrast, although usually cystic in the setting of ADPKD, will have mural soft tissue mass with thick septations with blood flow inside. In collaboration with CT scan, simple cysts appear as rounded structures with near water attenuation (0-15 HU). The wall is very thin and regular. Complex cyst mass which have had internal complications may be hyperdense with internal non-enhancing septations and/or calcifications & suspicious for malignant transformation. (5)

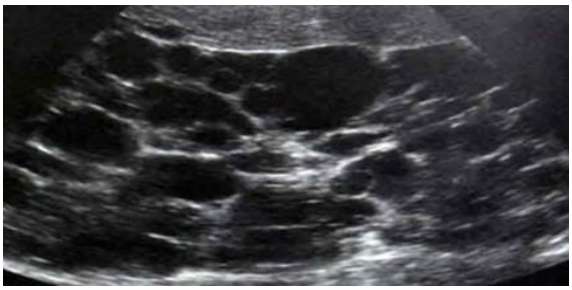

Figure (1): Abdominal ultrasound (oblique view) revealed markedly enlarged both kidneys, measured (22.5 X 12.5 cm) with multiple variable sized cysts.

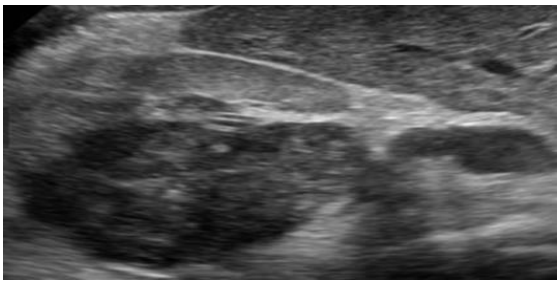

Figure (2): 2D abdominal ultrasound (transverse view) revealed a large irregular mass in the upper pole of right kidney, measured (8.5 X 7.5 cm) with heterogeneous echopattern.

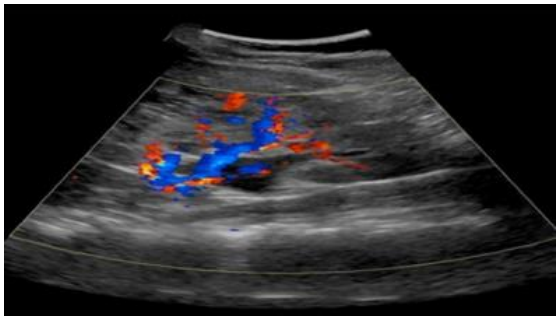

Figure (3): CCDI revealed abnormal internal vascularity in the right renal mass.

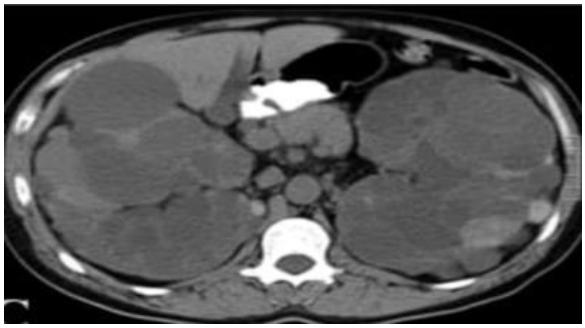

Figure (4): NCCT of abdomen revealed bilateral multiple variable sized renal cysts & the right kidney showed a soft tissue containing cystic lesion in the upper pole & thick internal septations impressing for renal cell carcinoma.

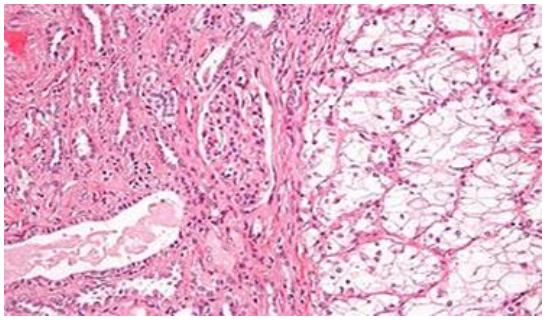

Figure (5): Histopathological examination of mass by H & E revealed clear cell type renal cell carcinoma.

Discussion:

The renal cell carcinoma (RCC) is a serious complication of autosomal dominant polycystic kidney disease. (6) Presence of hematuria owing to an underlying malignancy should prompt an urgent referral to urology for evaluation for nephrectomy.

(7) Radical right nephrectomy was done & histopathological study revealed renal cortical tumor characterized by malignant epithelial cells with clear cytoplasm and a compact-alveolar or acinar growth pattern interspersed with arborizing vasculature (Fig. 5). A variable proportion of cells with granular eosinophilic cytoplasm may be present. It is important to clarify the sub-type of renal cell carcinoma for accurate diagnosis & appropriate management.

Conclusion:

Sudden onset of gross hematuria in a case of ADPKD & end stage renal disease must raise the concern of malignant transformation of cystic lesion.

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