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A CASE OF ADPKD WITH ASSOCIATED EXTRA-RENAL ABNORMALITIES: INCIDENTALLY DETECTED.

Radiodiagnosis

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ABSTRACT

ADPKD, though uncommon, is important cause of progressive renal impairment leading to ESRD. It is a considered as a multisystem disease. Renal disease can be accompanied by abnormalities in other organs. Morbidity is determined by renal as well as extra-renal abnormalities. Here, we are presenting a case of ADPKD, detected on imaging studies, in a young adult male patient under evaluation for abdominal discomfort & episodic diarrhoea.

KEYWORDS

ADPKD, Seminal vesicle, Epididymis, Colonic Diverticula

INTRODUCTION:

ADPKD (Autosomal Dominant Polycystic Kidney Disease) is the most common renal cystic disease resulting from aberrations in gene. It usually manifests in young adults, is progressive & ultimately results in end stage renal disease (ESRD). Patients with ADPKD also have other abnormalities & morbidities i.e. extra-renal cysts (in liver, pancreas, seminal vesicles, epididymis, testis), cardiovascular abnormalities, & colonic diverticula. Early diagnosis of ADPKD & detection of other associated abnormalities is important as it will allow patient counseling, timely management & appropriate follow-up. Patient may come to notice because of clinical manifestation of extra-renal abnormalities.

Case Report:

A 24 Year Male patient was referred to Radiology department for CT abdomen in view of recurrent episodes of abdominal discomfort & diarrhoea. Non-contrast & contrast enhanced CT abdomen study was performed. CT revealed multiple small cortical cysts in both kidneys, more in left kidney, with few of left renal cysts being exophytic. There was no cyst wall calcification. Rest of bilateral kidney were normal. Renal cystic disease was not so obvious on ultrasound. CT also revealed bulky lobulated hypodense bilateral seminal vesicle, with USG correlation showing bulky seminal vesicles having cysts. Also noted on CT were few colonic diverticula – prominent in proximal ascending colon & distal transverse colon. No other obvious lesion was seen in colon. Stomach & small bowel loops were normal. Mild hepatomegaly was noted, however no focal liver lesion/cyst. Pancreas was normal. USG of scrotum revealed few small subcentimeter size cysts in epididymis, with normal appearing testes.

Though diverticula were missed on colonoscopy, there was no inflammatory change or polyp/mass in colon. Laboratory renal profile (S. Creatinine & Blood Urea) was normal. Haemogram was also normal. Patient was normotensive.

In view of multiple small cysts in bilateral kidney accompanied by seminal vesicle cysts, epididymal cysts & colonic diverticula, diagnosis of ADPKD was proposed.

DISCUSSION:

ADPKD usually manifests by age of 30 yrs & majority of patients develop ESRD by 60 years of age. Our patient was having no family history. However, number of cysts was more than five, even in a single kidney which is consistent for ADPKD as per Ravine's criteria for negative family history. [1]

There are no validated diagnostic CT criteria for ADPKD. However, CT has been used effectively in affected individuals with very mild disease when no cysts are detectable by US (5% of cases) – in our case renal cystic disease was less obvious on USG. Incidental discovery of renal cysts and PKD is most often made during the workup for abdominal pain. [2]

Seminal vesicle cysts have been reported in up to 60 % of cases of ADPKD. [3] In a article by Belet U et. al prevalence of seminal vesicle & epididymal cysts have been reported to be 39% & 18 % respectively. [4] In our case both seminal vesicles were bulky with cysts. Few small cysts were also noted in left epididymis. Cysts, however, are most frequent in liver. Cysts can be also seen in pancreas and spleen. In our case extra-renal cysts were seen in seminal vesicles & epididymis. No cysts were seen in liver, pancreas or spleen.

Colonic diverticula have been reported to be more in ADPKD patients with ESRD. In ADPKD without ESRD, prevalence of colonic diverticula has not been found to be significantly greater than age and sex matched general population. [5] In our case, patient was not having ESRD, however colonic diverticula were present.

Patients with colonic diverticula may be asymptomatic or may present with features of diverticulitis without inflammation (non-inflammatory diverticulitis) or with inflammation (acute/chronic diverticulitis). Patients with non-inflammatory diverticulitis do not show inflammatory changes in resected segments. Symptomatic patients may present with mild symptoms like abdominal discomfort & altered bowel habit or may present with pain, nausea, vomiting, fever, bleeding PR, & leukocytosis. [6] In our case, patient was having abdominal discomfort & episodic diarrhoea; however CT did not reveal any inflammatory change in bowel wall/diverticula. Colonoscopy also did not reveal any inflammatory change. This is consistent with non-inflammatory diverticulitis.

Acquired renal cystic disease was excluded as diagnosis because was patient was young, having normal renal function & normal kidney size. As cysts were mainly cortical & exophytic with no calcification, diagnosis of medullary cystic renal disease was also ruled-out. Patient in our case was not having any solid renal mass or clinical feature of Tuberous scleorsis. [7] VHL was ruled out as pancreas was normal, and there was neither solid renal lesion nor phaeochromocytoma, and also seminal vesicle cysts have not been mentioned as associated finding of VHL.

CONCLUSION:

Patient with ADPKD in early stage can present to clinician with non-specific clinical features. Radiologist while coming across a young or middle age patient with renal cysts with not classic picture of ADPKD, & normal renal function, should carefully look for ADPKD associated abnormalities, as with detection of such abnormalities diagnosis of ADPKD can be made with confidence, patient can be counseled regarding clinical scenario, and timely management can be started to control morbidity.

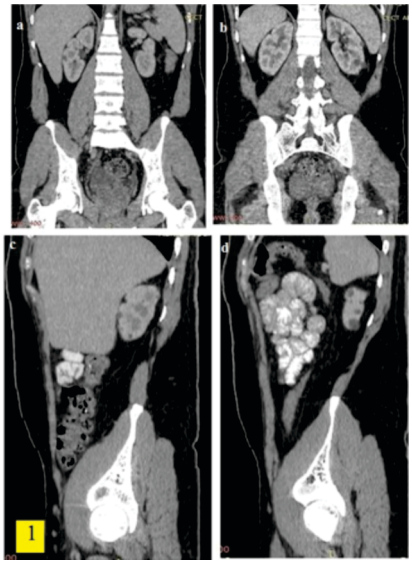
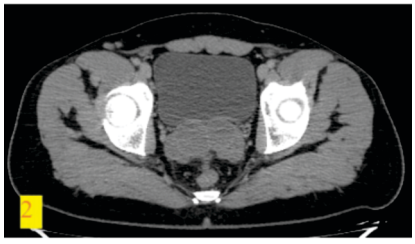


Figure-1: Coronal & sagittal reformatted CT images (a-d) showing multiple small cortical cysts in bilateral kidney. Also noted bulky lobulated hypodense seminal vesicles (a,b).



Fugure-2: Axial CT image through pelvis showing bulky lobulated hypodense seminal vesicles.

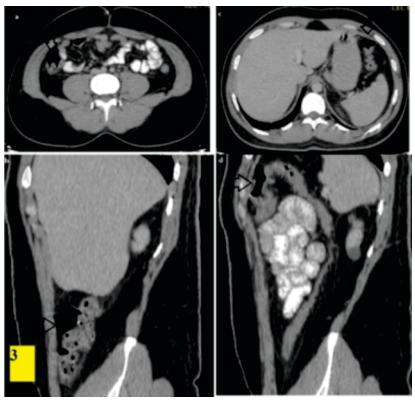


Figure-3: Axial CT images & sagittal reformats showing diverticula in ascending colon (a,b) & distal transverse colon (c,d).

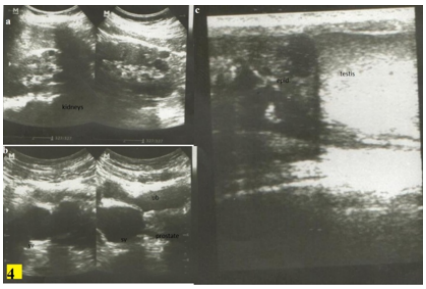


Figure-4: Ultrasound images showing cysts in bilateral kidney (a), seminal vesicles (b) & epididymis. Renal cystic disease was less obvious on ultrasound.

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