

Autosomal Dominant Polycystic Kidney Disease

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BRIEF HISTORY

A 28-year-old male presented with uncontrolled hypertension and vague abdominal discomfort for the past six months. On examination, his blood pressure was 184/96 mm of Hg and rest of his vitals were stable. Abdominal examination revealed a palpable liver and bilateral flank masses larger on the right side than the left side. The masses were bimanually palpable. His hemogram and routine biochemical panel were within normal limits. Contrast enhanced CT abdomen revealed multiple hepatic cysts and bilaterally enlarged kidneys with multiple cysts and nephrolithiasis (Figures 1 and 2). The diagnosis of polycystic disease of kidney and liver was made. Screening abdominal ultrasound revealed multiple family members with cystic renal disease making the diagnosis of autosomal dominant polycystic kidney disease (ADPKD) most likely. Genetic counseling was given and hypertension was controlled with a combination of antihypertensives which included enalapril, hydrochlorothiazide and amlodipine.

Hypertension in young patients often has a long list of differential diagnosis including ADPKD, which unlike most others, can often be diagnosed easily with a combination of bed side evaluation and simple abdominal imaging. ADPKD is the most common inherited renal disorder with an incidence of 1 in 400 to 1000 live births [1]. Although an inherited genetic disorder, the condition rarely presents in childhood and often presents in early adulthood. Mutations involving polycystic kidney disease (PKD) genes PKD1 and PKD2 constitute 85% and 15% of cases, respectively [2]. PKD1 gene on chromosome 16 codes for polycystin1, whereas PKD2 gene on chromosome 4 codes for polycystin2 [2]. The hallmark of the condition is the presence of enlarged kidneys with multiple cysts. Patients may also have cysts in liver, spleen, pancreas and the male reproductive tract [3]. Liver cysts have been demonstrated by imaging in up to 94% [4] patients by the age of 35 years; however, hepatic cysts tend to predominate in females [3, 4]. Other manifestations include cerebral aneurysms (5-7%) and colonic diverticula [3]. Abdominal pain is the most common complaint while systemic

Figure 1: Contrast CT abdomen showing multiple renal cysts of varying sizes. Renal calculus is visualized in right kidney (arrow head)



arterial hypertension is one of the earliest evidences of this genetic syndrome [5]. Ultrasound abdomen remains the initial modality for the diagnosis. Diagnostic criteria by Ravine et al requires that at risk individual should have at least two renal cysts (unilaterally or bilaterally) for the age group 15-29 years, at least two cysts in each kidney for 30-59 years and at least four cysts in each kidney for those ≥ 60 years [6]. However, this criterion often holds good only for PKD1 and is inaccurate for PKD2 cases. For at risk individuals irrespective of genotype, Pei et al criterion has been found to be superior to Ravine et al criteria [7]. According to Pei et al, for those with family history of ADPKD of unknown genotype diagnosis requires the following: in individuals between 15 and 39 years of age, the presence of three or more unilateral or bilateral kidney cysts; in individuals between 40 and 59 years of age, two or more cysts in each kidney; and in individuals ≥ 60 years of age, at least four cysts in each kidney [7]. CT, MRI and genotyping are also useful diagnostic modalities.

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Figure 2: Cysts in liver as well as in the upper pole of both kidneys



Patients often progress to develop end stage renal disease by fourth to sixth decade of life. So far no effective treatment is available for the syndrome and treatment is often tailored to the manifestations.

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