Severe autosomal dominant polycystic kidney disease

Neal S Gerstein,¹ Craig E Rumbaugh,¹ Julie M Riley²

¹Department of Anesthesiology and Critical Care Medicine, University of New Mexico School of Medicine, Albuquerque, New Mexico, USA

²Department of Surgery, University of New Mexico School of Medicine, Albuquerque, New Mexico, USA

Correspondence to
Dr Neal S Gerstein,
ngerstein@gmail.com

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DESCRIPTION

A 61-year-old man with a known history of autosomal dominant polycystic kidney disease (ADPKD) and stage IV chronic kidney disease presented with a 6-month history of abdominal pain, nausea, vomiting and fatigue. In addition to the ADPKD, the right kidney had a 4.4 cm inferior pole mass concerning for renal cell carcinoma (RCC). Preoperative imaging (figure 1), a coronal CT of abdomen and pelvis, demonstrates bilateral ADPKD. His total kidney volume was calculated to be 9980.5 mL, which in combination with his age made him a '1E' (most severe) based on the Mayo Clinic risk stratification schema. The following aggregate of issues led the patient to undergo bilateral open nephrectomies: the suspicion for malignancy associated with the right renal mass, a slight increase in malignancy risk associated with polycystic kidney disease in general, and the likely need for postoperative dialysis if left with only a single poorly function kidney in conjunction with a desire to be placed on the renal transplant list (see figure 2 demonstrating excised gross specimens).

ADPKD is a relatively common disease with a prevalence of 1:400–1:1000 and is associated with up to 10% of end-stage renal disease aetiology. However, due to age-dependent penetrance and

Figure 2 Gross surgical specimens: (A) right kidney and (B) left kidney.

variable expressivity, a minority of patients may go undiagnosed throughout their lifetime.¹ Although there is extensive allele heterogeneity, the ADPKD-causing mutations are found in the *PKD1* locus (chromosome 16 abnormality (16.p13.3); 78% of cases), the *PKD2* locus (chromosome 4 abnormality (4p21); 15% of cases), and a rarer and newly discovered third locus, *GANAB* (chromosome 11 abnormality (11q12.3); 0.3% of cases); the *GANAB* locus is also associated with autosomal dominant polycystic liver disease.²

The age of renal failure onset is variable but typically occurs by the fourth to sixth decade of life. The most common presentation is a urinary tract infection.¹ Flank or abdominal pain is also a common presenting symptom and may be due to renal capsule expansion, renal pedicle traction, compression of adjacent abdominal organs, nephrolithiasis, renal haemorrhage and chronic urinary

Figure 1 Coronal CT of abdomen and pelvis. Image demonstrates bilateral renal parenchyma that is diffusely replaced by innumerable cysts (arrows), a few of which are hyperdense. There is no hydronephrosis. There are numerous hepatic cysts (star).

Learning points

- ▶ Autosomal dominant polycystic kidney disease is a relatively common disease with a prevalence of 1:400–1:1000 and the most common genetic renal disease in adults. However, due to age-dependent penetrance and variable expressivity, a minority of patients may go undiagnosed throughout their lifetime.
- ▶ The age of renal failure onset is variable but typically occurs by the fourth to sixth decade of life.
- ▶ The most common symptoms are flank or abdominal pain, along with hypertension and haematuria. Diagnosis relies on family history, imaging with typical findings of enlarged kidneys containing multiple cysts, and potentially genetic testing.

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tract infections.¹ Diagnosis relies on imaging with typical findings of enlarged kidneys containing multiple cysts. Although less reliable in younger patients (less than 18 years), screening renal ultrasonography should be considered in asymptomatic patients with an ADPKD family history; moreover, some centres mandate genetic testing for a definitive diagnosis.¹

Other extrarenal manifestations of ADPKD include hypertension (occurring in 60% of patients with ADPKD prior to renal function impairment), cerebral aneurysms, hepatic cysts, pancreatic cysts, cardiac valvular disease (mitral, aortic and tricuspid insufficiency), aortic root dilation, colonic diverticula, abdominal wall and inguinal hernias, and seminal vesicle cysts.^{1,3} Lastly, there is a reported association between RCC and ADPKD. RCC may present as a cystic renal mass, and a small percentage of electively excised polycystic kidneys are found to contain a malignant neoplasm. RCC should be considered if a complex cyst (defined as a cyst with calcifications, septations or wall thickening) grows rapidly or is associated with symptoms that are inconsistent with the underlying disease severity.¹

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