key points

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Polycystic kidney disease (PKD) is the most common cause of inherited kidney disease worldwide. It has a prevalence of < 5 per 10,000 population and accounts for 7% of cases of end-stage renal disease (ESRD) in the UK. PKD is a genetic disorder with an autosomal dominant mode of inheritance; hence any child of an affected parent has a 50% chance of inheriting the condition. Despite this 25% of patients will have no known family history and these cases are thought to be due to de novo mutations in the causative genes. Routine genetic testing is often unnecessary in those with a family history of PKD. Diagnosis is primarily made on the basis of a positive family history plus an age-specific minimum number of cysts on ultrasonography.

PKD can present asymptomatically and should be considered in any patient with renal cysts noted on abdominal imaging, especially if there is a history of hypertension. Patients with PKD may present with hypertension before a decline in eGFR is noted. As PKD progresses there is gradual loss of kidney function, with a predictable decrease in eGFR.

Cyst haemorrhage, rupture or infection can all present with acute loin pain. Infections can be severe and associated with systemic upset and sepsis. Bleeding can occur into cysts and as a result may not be 'visible' but can be sufficiently severe to cause symptomatic anaemia. Visible haematuria can occur in the setting of cyst rupture or haemorrhage if the involved cyst communicates with the urinary tract.

PKD can cause a number of extrarenal complications. Liver cysts affect around 80% of PKD patients and can cause liver capsular pain, early satiety, nausea and vomiting. Intracranial haemorrhage due to rupture of an intracranial berry aneurysm is a potentially life-threatening complication of PKD and should be considered in any patient presenting with thunderclap headache, new focal neurology or unexplained syncope. Mitral valve prolapse is common and other valvular and aortic abnormalities can occur.

Angiotensin converting enzyme inhibitors or angiotensin receptor blockers are the first choice antihypertensive agents in PKD. The target clinic blood pressure should be < 130/80 mmHg. Lipid-soluble antibiotics, such as ciprofloxacin, are effective for cyst infections as they achieve better cyst penetration than non-lipid soluble agents. Tolvaptan, a vasopressin receptor antagonist, can be used in some PKD patients and can slow cyst development and the rate of decline in renal function.

The median age at onset of ESRD is 54 years for patients with a PKD1 mutation compared with 69 years for patients with a PKD2 mutation. Planning for renal replacement therapy (kidney transplant or dialysis) should start well in advance of the onset of ESRD.