key points

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Polycystic kidney disease (PKD) is a common hereditary disorder. It is characterised by multiple renal cysts, slowly enlarging kidneys and a gradual decline in renal function, which results in end-stage renal disease (ESRD) in the majority of patients by the fifth to sixth decade of life. A child of an affected individual has a 50% chance of inheriting the disorder.

PKD is a multisystem disorder with a variety of renal and non-renal manifestations. Renal features of PKD occur as a direct consequence of the cysts and the progressive renal failure they induce. Hypertension precedes the onset of renal failure in the majority of patients and is almost universally present in those with established renal failure. Left ventricular hypertrophy is commonly associated with hypertension. Loin pain occurs with enlarging cysts, cyst infection or haemorrhage and renal stones. Chronic severe pain is rare and an alternative explanation should be sought. Gross haematuria occurs in up to 40% of patients. Infection is common and can present as cystitis, pyelonephritis, cyst infection, or rarely, perinephric abscess.

Subarachnoid haemorrhage resulting from a ruptured berry aneurysm is one of the most devastating presentations of PKD and can result in death or severe disability. Polycystic liver disease is the most common non-renal manifestation of PKD. Mitral valve prolapse is the most common valvular abnormality associated with PKD and occurs in up to 25% of patients.

An abdominal ultrasound scan can detect cysts more than 1cm in diameter. If a patient has a family history of PKD, the following criteria are used to establish diagnosis:

Positive family history + patient age:

- <30 years: at least two cysts (unilateral or bilateral)
- 30-59 years: at least two cysts in each kidney
- >60 years: at least four cysts in each kidney.

Reasons for referral to specialist nephrology services include: to confirm PKD and discuss potential screening of family members; to manage complications, including poorly controlled hypertension, frequent urinary tract or cyst infections and recurrent renal calculi; if the patient is pregnant or planning a pregnancy; and to manage progressive renal failure and prepare for ESRD.

Patients with PKD and early chronic kidney disease (CKD) can be managed in primary care in accordance with CKD guidelines. Patients should have a biannual assessment that includes urinalysis, blood pressure, renal function (serum creatinine and estimated GFR), protein estimation (albumin or protein:creatinine ratio) and cardiovascular risk. Control of blood pressure is arguably the most important management goal. Patients with PKD should have a target blood pressure of 125/75 mmHg.