key points

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Polycystic kidney disease (PKD) is the most common inherited renal disorder that results in chronic kidney disease. PKD has an autosomal dominant pattern of inheritance. The prevalence is between 1:500 and 1:1,000. Up to 10% of adults with end-stage renal disease (ESRD) have a genetic disorder such as PKD. A family history of PKD may be absent in up to 25% of affected individuals. The most common clinical features are visible haematuria, loin pain, UTI and hypertension.

The typical clinical course is a progressive increase in the number and size of renal cysts associated with gradual loss of kidney function (falling eGFR). Risk factors for progression include: younger age at diagnosis; large kidney volume; rapid cyst growth; hypertension; male gender; and visible haematuria. Approximately 50% of individuals with PKD will require renal replacement therapy by the sixth decade of life.

PKD is a multisystem disorder associated with multiple bilateral renal cysts, slowly increasing kidney size and progressive chronic kidney disease. A diagnosis of PKD is confirmed by ultrasound scan showing the presence of multiple kidney cysts. More than 80% will also have multiple liver cysts, which can lead to local pressure effects. Cerebral haemorrhage, secondary to rupture of a berry aneurysm, occurs in up to 8% of individuals. Mitral valve prolapse occurs in up to 25% of patients. Aortic and tricuspid valve prolapse, along with aortic root dilation and ascending aortic aneurysm and dissection have all been reported in patients with PKD.

Excellent control of blood pressure is a key element in management, < 130/80 mmHg is a reasonable target using angiotensin converting enzyme inhibitor or angiotensin receptor blocker therapy. Calcium channel blockers can be added. Diuretics should not be prescribed because they are associated with fluid volume depletion and stimulation of ADH secretion. Higher levels of ADH are associated with more rapid loss of renal function.

Several drugs designed to limit cyst growth have shown promise in clinical trials (tolvaptan, octreotide, everolimus). NICE has approved the use of tolvaptan to slow the progression of cyst development in adults with PKD who have chronic kidney disease stage 2 or 3 with evidence of rapidly progressive disease. Renal replacement therapy, dialysis and transplantation, are treatment options for ESRD. Potential living related kidney donors need to be screened carefully to ensure that they do not have PKD.