key points

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Polycystic kidney disease and Alport's syndrome are the most common causes of inherited renal disease in the UK. An average GP practice is likely to have at least six patients with autosomal dominant polycystic kidney disease (ADPKD). The disorder is characterised by the formation of fluid-filled cysts in the kidneys resulting in progressive renal impairment. Mutations in two genes have been identified. The PKD1 gene abnormality is responsible for 85% of cases of ADPKD, patients with PKD2 mutations typically present later and progress more slowly.

Patients with ADPKD can present with a positive family history, hypertension, flank pain, haematuria, renal insufficiency or proteinuria. The diagnosis has traditionally been based on ultrasound imaging. In at-risk individuals aged 15-39 years old, ≥ 3 cysts unilateral or bilateral is diagnostic. Genetic testing is rarely carried out as the genes are very large, extremely intricate, and ADPKD can be caused by different mutations at the same locus.

Screening will reduce the incidence of a late diagnosis when renal disease is advanced but a normal ultrasound scan in those under 30 years old is not conclusive. It is not recommended that children are screened. Once individuals are 18 years old, they can make an informed choice. Progression to end-stage renal disease (ESRD) is not inevitable in PKD, particularly in those with milder genetic mutations. The key to minimising the rate of progressive disease is tight blood pressure control. ACE inhibitors are recommended as the initial antihypertensive agent unless contraindicated.

Alport's syndrome is a disorder characterised by abnormal type IV collagen which is found in the kidney, eyes, skin and ears. Only around one in ten practices are likely to have a patient with Alport's syndrome. Eighty per cent of patients have the X-linked form of the disease. Men typically have a more severe phenotype with 50% of males reaching ESRD by the age of 25 and 100% by the age of 40. All first-degree relatives of a patient with confirmed Alport's syndrome should be offered screening. The combination of reduced hearing and urinary abnormalities in a young boy should alert GPs to consider this as a possible diagnosis and initiate referral. Diagnosis can be confirmed by renal or skin biopsy.

Thin basement membrane disease is also caused by an abnormality of type IV collagen but is a benign condition in the vast majority of patients. The inheritance pattern is autosomal dominant. Typically the presentation is with non-visible haematuria detected during routine health screening, potentially with a family history of haematuria but not renal failure.