

Autosomal dominant polycystic kidney disease

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Autosomal dominant polycystic kidney disease (ADPKD) is the most common autosomal dominant genetic cystic renal disease causing chronic renal failure requiring dialysis and renal transplantation. It occurs as a result of mutation in one of two genes (PKD1 on chromosome 16 and PKD2 on chromosome 4). ADPKD gene loci can be identified in individuals with a family history before the development of cysts begins; this is helpful in screening a potential sibling for kidney donation. ADPKD has variable penetration and approximately 50% of affected individuals eventually develop end-stage renal disease (ESRD). Risk factors for the development of ESRD are: /uni25CF early age of presentation; /uni25CF hypertension; /uni25CF male sex; /uni25CF ADPKD gene 1; /uni25CF African ethnic group. ADPKD is associated with cysts in other organs, such as the liver, pancreas, arachnoid membranes and seminal vesicles. It does not usually manifest before the age of 30 years and in some patients it is never diagnosed. Renal symptoms include abdominal pain, haematuria or a palpable mass. Most patients older than 20 years are hypertensive and good control of blood pressure can delay progression to renal failure. Novel agents such as vasopressin antagonists (tolvaptan), somatostatin analogues and mammalian target of rapamycin (mTOR) inhibitors have shown potential to prevent cystogenesis, cyst expansion and declining renal function. Intracranial aneurysms occur in approximately 10–30% of patients with ADPKD and subarachnoid haemorrhage may cause sudden death in young adults. Summary box 82.2 Renal cystic disease /uni25CF - /uni25CF /uni25CF /uni25CF /uni25CF

Double 'J' stent Lateral spatulation of the ureteral end Bosniak renal cyst classification is used to grade cysts and probability of malignancy ADPKD – autosomal dominant, systemic disease: Rarely manifests before the fourth decade Hypertension, abdominal pain, haematuria or a palpable /f_lank mass are common presentations Control of hypertension can delay progression

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