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1: Nephrology (Carlton). 2006 Apr;11(2):124-30. Related Articles, Links

Optimal care of autosomal dominant polycystic kidney disease patients (Review Article).

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SUMMARY: Autosomal dominant polycystic kidney disease (ADPKD) is the most common life-threatening, hereditary disease. The prevalence of ADPKD is more common than Huntington disease, haemophilia, sickle cell disease, cystic fibrosis, myotonic dystrophy and Down syndrome combined. In recent years there have not only been advances in the understanding of the genetic and molecular events involved in ADPKD, but some diagnostic and therapeutic advances have also emerged. In the genetics area, the gene for PKD1 was localised to chromosome 16, is associated with polycystin-2 protein, and found to account for approximately 85% of patients with ADPKD. The gene for PKD2, found in chromosome 4, accounts for approximately 15% of ADPKD, and is associated with the polycystin-2 protein. While these genetic and molecular biology findings have stimulated a great deal of exciting basic research in ADPKD, therapies to decrease morbidity and mortality in ADPKD patients have yet to emerge from these findings. In contrast, the early diagnosis and treatment of hypertension with inhibitors of the renin-angiotensin-aldosterone system have the potential to decrease or prevent left ventricular hypertrophy cardiac complications and slow the progression of the renal disease.

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