Autosomal dominant polycystic kidney disease

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1 Autosomal dominant polycystic kidney disease affects about 1 in 1000 people

Mutations in *PKD1* or *PKD2* cause autosomal dominant polycystic kidney disease, which proceeds to kidney failure in 70% of patients between the fourth and seventh decade of life.[1]

2 Autosomal dominant polycystic kidney disease is diagnosed by ultrasound

Signs of autosomal dominant polycystic kidney disease (i.e., numerous cysts and enlargement of the kidneys) may be seen on ultrasound performed in patients with abdominal pain, hematuria, early-onset hypertension or a family history of the disease. In people with a family history, the diagnosis may be made if three or more cysts are seen bilaterally on ultrasound in those aged 15 to 39 years, and with four or more cysts bilaterally in older patients. [2]

3 Supportive care includes management of hypertension, infection and pain

Inhibition of the renin–angiotensin–aldosterone system and aggressive blood pressure control (< 110/75 mm Hg) should be considered in patients with autosomal dominant polycystic kidney disease who are younger than 50 years and have preserved kidney function.[3] Cyst infections should be treated with cyst-penetrating antibiotics (i.e., fluoroquinolone) for a minimum of two to four weeks, and possible cyst drainage.[4] Chronic abdominal discomfort is common and acute-on-chronic pain secondary to cyst hemorrhage or nephrolithiasis can occur.[4]

4 About 5% of patients will have asymptomatic intracranial aneurysms

Screening of asymptomatic patients is not cost-effective. According to expert opinion, magnetic resonance imaging (MRI) is recommended only in those with family history of intracranial aneurysm or subarachnoid hemorrhage or a previous aneurysm rupture or those with high-risk professions (i.e., pilots).[4]

5 Patients with autosomal dominant polycystic kidney disease should have early referral to a nephrologist

Treatment should be started early, before loss of kidney function.[4,5] Total kidney volume assessed by MRI increases before appreciable loss of kidney function and is a valuable risk stratification tool. Genetic testing is helpful for diagnosis in select cases. Canadian consensus guidelines suggest that nephrologists consider tolvaptan, an antidiuretic hormone V2 receptor antagonist, in high-risk patients (Appendix 1, available at www.cmaj.ca/lookup/suppl/doi:10.1503/cmaj.170443/-/DC1) with preserved kidney function, with benefit balanced by cost, aquaretic adverse effects and potential hepatotoxicity.[3]

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