What Is Autosomal Dominant Polycystic Kidney Disease?

Autosomal dominant polycystic kidney disease is the most common genetic cause of kidney failure.

Autosomal dominant polycystic kidney disease (ADPKD) is a genetic disease in which individuals have a 50% chance of inheriting an ADPKD gene variant if one of their parents has ADPKD. However, in about 15% of patients, ADPKD arises from a genetic variant that was not inherited from either parent. ADPKD affects between 1 in 1000 and 1 in 2500 people worldwide and involves progressive growth of fluid-filled cysts in the kidneys.

People with ADPKD may also have cysts in their liver and pancreas, abdominal hernias, and cardiac valve abnormalities. Between 9% and 12% of patients with ADPKD have brain blood vessel abnormalities (aneurysms). Very rarely, these brain aneurysms can burst, which may result in permanent neurological damage or death.

Common Signs and Symptoms of ADPKD

In the early stages of disease, many individuals with ADPKD have no symptoms, although most have high blood pressure. As the kidney cysts increase in size, patients may develop abdominal pain or pressure, blood in the urine, kidney infections, and kidney stones. Over time, more than half of people with ADPKD develop kidney failure and need treatment with dialysis or kidney transplant. Onset of kidney failure is usually after age 50 years but may occur earlier.

Diagnosis of ADPKD

The recommended imaging test to diagnose ADPKD is ultrasound, which reveals cysts in the kidneys. Other imaging tests that may be considered are magnetic resonance imaging (MRI) or contrast-enhanced computed tomography (CT). Presence of kidney cysts confirms the diagnosis of ADPKD in people with a family history of ADPKD. If the diagnosis of ADPKD is uncertain based on imaging, genetic testing should be performed to confirm the diagnosis.

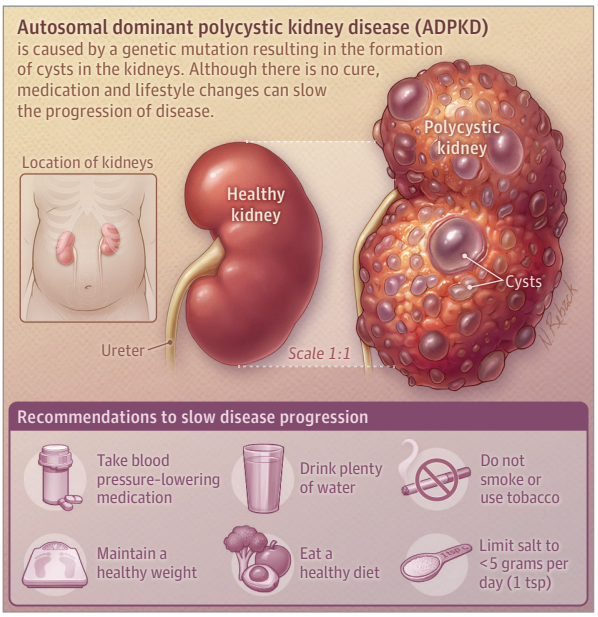
ADPKD Factors for Higher Risk of Kidney Failure

In ADPKD, certain categories of genetic variants are associated with more mild or severe disease. However, people with the same genetic variant may have different rates of progression to kidney failure. Kidney volume (adjusted for height and age) may help determine whether people with ADPKD are at a high risk of kidney failure because enlarged kidneys are associated with loss of kidney function.

Treatment of and Screening for ADPKD

Although there is currently no cure for ADPKD, treatments are available to improve outcomes. Medications to decrease blood pressure help slow the progression of kidney disease. Individuals with ADPKD should maintain an optimal weight, engage in regular physical exercise, have high water intake, limit salt to 5 grams per day, avoid smoking, and limit use of nonsteroidal anti-inflammatory drugs such as ibuprofen. A medication called tolvaptan may be taken by patients with ADPKD who are at higher risk of kidney failure but should be prescribed only by a physician experienced with this treatment.

Asymptomatic children of a parent with ADPKD should be closely monitored for high blood pressure but do not require screening before adulthood. Prior to being tested for ADPKD, adults with a family history of ADPKD should receive diagnostic counseling about the benefits of screening (timely initiation of treatment if ADPKD is diagnosed or reassurance if ADPKD is ruled out) and risks (potential problems with insurance and psychological aspects of being diagnosed with a genetic disease).



FOR MORE INFORMATION

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