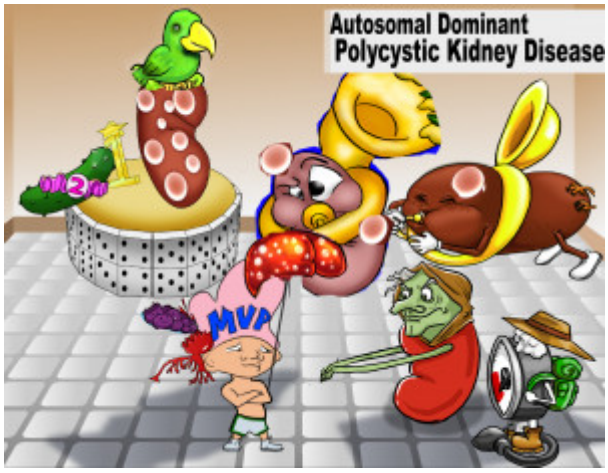
Autosomal Dominant Polycystic Kidney Disease

Autosomal dominant polycystic kidney disease (ADPKD) is an autosomal dominant condition. The majority of cases are thought to result from genetic mutations in the PDK1 and PKD2 genes which are located on chromosomes 16 and 4, respectively. The disease is characterized by cystic enlargement of the renal tubules. Patients are generally asymptomatic early in the disease, with the most common initial presenting symptom being hypertension. Nearly all patients with ADPKD experience progressive deterioration in renal function leading to chronic kidney disease. Patients are also at risk for various extrarenal manifestations including liver cysts, mitral valve prolapse, and intracranial aneurysms. Nephroprotective agents like ACE inhibitors or ARBs are helpful but many patients will require hemodialysis.



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Characteristics

Autosomal Dominant

Dominoes

As its name implies, autosomal dominant polycystic kidney disease is inherited in an autosomal dominant fashion. This means an individual need only inherit the mutated allele from one parent in order to manifest the disease.

Mutations in PKD1/2

PicKleD (1)-wand and (2)-tutu

Most cases of ADPKD are related to mutations of the PKD1 or PKD2 genes. These genes encode for proteins that regulate the differentiation and proliferation of renal tubular epithelial cells. The two loci segregate independently as they are located on separate chromosomes, with the PKD1 and PKD2 genes residing on chromosomes 16 and 4, respectively.

Cystic Enlargement of Renal Tubules

Cysts on Enlarged Kidneys with Tubas

ADPKD is characterized by cystic enlargement of renal tubules. It is thought that the mutations in PKD1/2 lead to dysregulation of the proliferation and differentiation of renal tubular epithelial cells which has been proposed to play a role, but the exact mechanism of how this leads to cyst formation is not fully understood. The cystic enlargement can be readily appreciated on ultrasound.

Clinical Features

Chronic Kidney Disease

Crone Kidney Diseased

ADPKD is the leading genetic cause of end-stage renal disease (ESRD) and is the fourth overall cause of ESRD. Over the course of their lifetimes, patients will experience progressive renal failure related to enlargement of tubular cysts. Patients with ADPKD have a >50% chance of progressing to ESRD by age 70.

Hypertension

Hiker-BP

Early polycystic kidney disease is typically asymptomatic, and diagnosis is therefore often delayed in patients with no known family history of ADPKD. Hypertension is most commonly one of the first presenting signs, and is seen in 40-50% of patients on initial presentation.

Mitral Valve Prolapse

Mitt-troll MVP

Cardiac valve abnormalities are common in patients with ADPKD. Several studies have found roughly 25% of patients with ADPKD have ultrasound studies consistent with mitral valve prolapse. Other cardiac abnormalities such as aortic insufficiency, tricuspid valve prolapse, and mitral insufficiency are also seen to a higher degree in these patients than in the general population, but mitral valve prolapse is the most common.

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Hepatic Cysts

Liver-balloon with Cysts

Patients with ADPKD are likely to form cysts in locations other than the kidneys. The most common extrarenal site of cyst formation is the liver, as over 80% of patients with ADPKD have been shown to develop hepatic cysts at some point over the course of their lifetime. Pancreatic, ovarian, and splenic cysts may also be seen.

Berry (Saccular) Aneurysms

Berries and Bulging-aneurysm

Patients with ADPKD are at an increased risk of developing intracranial berry (or saccular) aneurysms, which have been found to be present in as many as 5-10% of patients with ADPKD. There is a strong familial pattern to this phenomenon, and patients should be screened with MRI, as failure to recognize or manage an aneurysm could result in stroke or death.

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