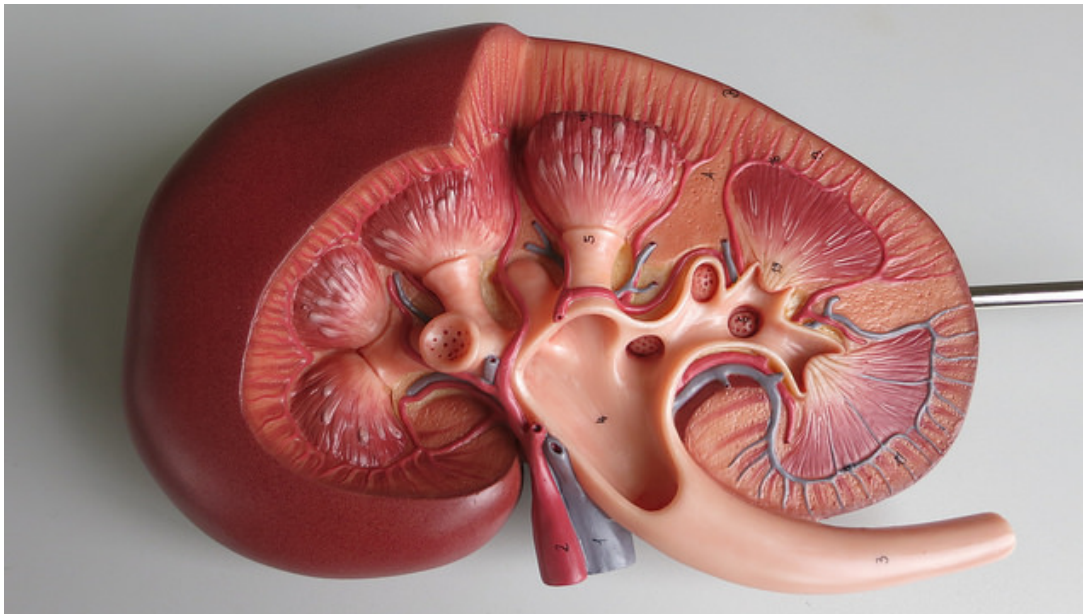


Types of Kidney Cysts (Renal Cysts) — Diagnosis and Treatment

[See online here](#)

A kidney cyst can arise in any part of the nephron and collecting ducts, and can be found incidentally on imaging tests, or can be part of a renal or systemic disease. Renal cystic diseases consist of a large spectrum of diseases that differ in regards to pathophysiology, prognosis and treatment and that can be usefully divided into hereditary, acquired and developmental conditions. Hereditary renal cystic diseases often lead to kidney failure and are commonly associated with extrarenal manifestations, whereas non-genetic cystic disorders are usually limited to the kidney and rarely result in kidney functional deterioration.



Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Epidemiology of ADPKD

ADPKD is the most commonly inherited renal disease and affects approximately 1 in 800 live births. ADPKD is also responsible for 6-10% of renal therapy replacement treatments and is the leading [genetic](#) cause of end stage renal disease worldwide.

Etiology and Pathophysiology of ADPKD



Image: "Gross Pathology of Polycystic Kidneys" by Patho. License: Public Domain

Two mutations, which are inherited in an autosomal dominant fashion, are responsible for the disease: **PKD1 (chromosome 16)** in 85% of cases and **PKD2 (chromosome 4)** in 15% of cases. Nonetheless, the condition has a variable phenotypic expression which can be explained by the two-hit hypothesis, in which a second somatic mutation (second hit) appears in the normal allele and leads ultimately to cell proliferation and transverse growth, resulting in the formation of cysts that involve all parts of the nephron in the cortex and medulla.

Growth of **renal cysts** causes **growth and deformation of the kidney, tubular obstruction** and **renal ischemia**, leading to **increased renin-angiotensin-aldosterone activity and hypertension**. **Kidney** function progressively deteriorates, but does not decline until at least 50% of the parenchyma is destroyed.

Clinical Presentation and Symptoms of ADPKD

Abdominal or flank pain is a prominent presenting symptom in patients with ADPKD and is almost universally present. Patients also commonly complain of fatigue, breathlessness, weakness and malaise in the early stages of the disease. **Hematuria** is another frequent presenting manifestation, but is usually self-limited and lasts approximately one week. On physical exam, patients commonly have **diastolic hypertension**, which is notable for becoming less problematic as kidney function further deteriorates.

Additional findings include **palpable bilateral flank masses**. Symptoms that are common with kidney failure such as **edema and pallor** are rare on initial presentation. Extrarenal manifestations include **benign cysts in the liver** (94%), the seminal vesicle (40%) and the pancreas (9%), as well as connective tissue abnormalities such as **colonic diverticula**, **mitral valve prolapse** (25%), **intracranial aneurysms** (8%) and abdominal hernia (10%).

Laboratory Evaluation and Diagnosis of ADPKD

Ultrasound is the diagnostic modality of choice for ADPKD. It should also be used for **screening family members**. Other possible diagnostic imaging tests include MRA, MRI and CT-scans. The disease may also be associated with **increased hematocrit** due to elevated **erythropoietin** secretion from the cysts.

Microalbuminuria is also common but does not reach nephrotic-range proteinuria. Select patients with ADPKD should undergo **screening for intracranial aneurysms**. Indications for screening include family history of intracranial aneurysms and/or bleeding, new-onset severe headache, major elective surgery and CNS signs and symptoms.



Image: "Abdominal CT scan of an adult with autosomal dominant polycystic kidney disease. Extensive cyst formation is seen over both kidneys, with a few cysts in the liver as well." by Steven Fruitsmaak. License: [CC BY-SA 3.0](https://creativecommons.org/licenses/by-sa/3.0/)

Therapy and Treatment of ADPKD

Strict blood pressure control is fundamental with either an ACE inhibitor or an ARB. Clinical studies found **Vasopressin V2 receptor antagonists**, such as **tolvaptan**, to be useful. Regular monitoring of the kidney parameters, as well as ultrasound controls, play an important part in the management of the patient.

Renal replacement therapy (dialysis) is usually required for 50% of patients above the age of 60. **Nephrectomy** is reserved for cases of **anatomical infringement, cyst hemorrhage, infection and uncontrolled pain**. A **kidney transplant** is the treatment of choice for patients with **end-stage renal disease**.

Prognosis for ADPKD

50 to 75% of patients with ADPKD will require dialysis by the age of 75. Some predictors of rapid deterioration include PKD1 mutation, large kidney size, proteinuria, male sex, and early age at diagnosis. There is **no concomitant increase in renal cancer**.

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

Epidemiology of ARPKD

ARPKD is a **childhood** kidney disease with incidence of about 1/10.000 to 1/20.000 birth.

It affects all racial and ethnic groups and males and females equally.

Etiology and Pathophysiology of ARPKD



Image: "Baby with distended abdomen due to voluminous kidneys" by Carsten Bergmann, Pediatric Nephrology. License: Open Access

ARPKD is caused by a mutation in the **PKHD1 gene** on the short arm of **chromosome 6**. The protein encoded by the gene is expressed by renal and hepatic epithelial cells, but its function remains only partially understood. ARPKD leads to **bilateral, non-obstructive** elongation of the collecting ducts, resulting ultimately in **enlarged kidneys**. All patients will also have **congenital hepatic fibrosis**, which can be more severe than the renal disease. It can lead to **portal hypertension** and ultimately **GI hemorrhage, varices and splenomegaly**.

Clinical Presentation and Symptoms of ARPKD



Image: "Potter's phenotype with distinctive facial features" by Carsten Bergmann, Pediatric Nephrology. License: Open Access

Patients with ARPKD might present **at birth** with enlarged **flank masses** that may complicate delivery. These babies will also have **Potter facies** (parrot beak nose, low-set

ears) and abnormal limbs. Older infants may present with **abdominal distention** due to renal masses or hepatosplenomegaly. **Hypertension** can be severe and can be the presenting symptom.

Laboratory Evaluation and Diagnosis of ARPKD

Ultrasonography is the primary diagnostic method, particularly among neonates, although a definitive diagnosis sometimes may require a **biopsy**. **Genetic testing** can also be used when clinical criteria are not met. Renin levels are usually normal.

Therapy and Treatment of ARPKD

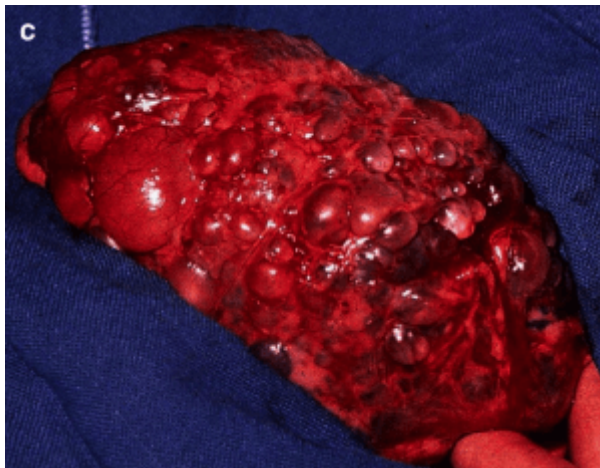


Image: "Macroscopic appearance of advanced-stage autosomal dominant polycystic kidney disease (ADPKD) showing enlarged kidneys with multiple cysts that almost completely destroyed and replaced the renal parenchyma" by Carsten Bergmann, Pediatric Nephrology. License: Open Access

Neonatal survival is dependent on ventilation and intensive care unit treatment. Hypertension needs to be treated preferably with **ACE inhibitors**. Once chronic kidney disease develops, patients may require iron supplements and erythropoietin for anemia, calcium, phosphate binders and suppressors of the parathyroid to prevent metabolic bone disease, and growth hormone to fend off the effects of uremia on growth.

Unilateral or bilateral nephrectomy is performed in cases of respiratory compromise in neonates or when failure to thrive is present. **Dialysis and transplantation** are treatments of choice in **end-stage renal disease**.

Prognosis for ARPKD

Prognosis varies considerably. Patients who are born with **oligohydramnios** most often die due to pulmonary complications. Patients who survive the neonatal period can still develop chronic kidney disease, although prognosis has improved due to renal transplants. **Congenital hepatic fibrosis** can still lead to significant morbidity.

Medullary Cystic Kidney Disease (MCK)

Epidemiology of MCK

MCK is a very rare genetic disorder that has been mostly reported in the United States; both genders are equally affected with no racial predilection.

Etiology and Pathophysiology of MCK

MCK presents in two types, and both are inherited in an **autosomal dominant** fashion. The gene responsible for type 1 is localized to **chromosome 1**, whereas the gene responsible for type 2 is localized on **chromosome 16**. Nonetheless, 16% of patients have no family history, suggesting a sporadic mutation.

The disease results in the formation of cysts in the medulla and typically presents in **adulthood**. These cysts may not be detected and may result in progressive kidney failure and a **decrease in kidney size**. Pathophysiology includes **reduced urinary concentration capacity** and **loss of sodium conservation** leading inevitably to **renal failure**.

Histological findings include **uniformal thinning of renal cortex** and a **segmental distribution of cysts** of varying sizes in the **medulla** and **corticomedullary junction**. The presence of cysts is a late finding and may not be found on a biopsy.

Clinical Presentation and Symptoms of MCK

Patients present with **polyuria and polydipsia** due to a reduced urinary concentration capacity. Median onset age for type 1 disease is 62 years and for type 2 is 32 years. Extrarenal manifestations are limited to **hyperuricemia and gout**. **Anemia** is common and may present before renal disease.

Laboratory Evaluation and Diagnosis of MCK

Urinalysis can be helpful and shows **low specific gravity**. **Proteinuria** is usually mild. The diagnostic study of choice is **thin-section CT with contrast**. It will show cysts in the medulla and corticomedullary junction. **Ultrasonography** is also helpful and will show normal or a moderate reduction in size with a loss of corticomedullary differentiation.

Therapy and Treatment of MCK

End-stage renal disease develops in all cases and management is symptomatic. **Erythropoietin** is the treatment of choice for associated anemia. Eventually, patients will require **dialysis** and **renal transplants**.

Prognosis for MCK

Eventually, all patients develop end-stage renal disease.

Multicystic Dysplastic Kidney (MCDK)

Epidemiology of MCDK

MCDK is one of the most frequent **congenital kidney disorders**. Unilateral MCDK occurs in 1 of 4300 live births and cumulative incidence of unilateral and bilateral occurs

in 1 of 3600 live births. Involvement of the left kidney occurs in 55% of cases, whereas, that of the right kidney, occurs in 45%.

Etiology and Pathophysiology of MCDK

MCDK is mostly a sporadic disease, although familial inheritance has been reported. It is due to dysfunction in the early developmental period and is due to an abnormal induction of the **metanephric mesenchyme** (which later becomes kidney tissue) with the ureteral bud. Both exposure to viral infections, as well as spontaneous genetic mutations, have been shown to be involved in the pathogenesis.

MCDK may progress with no change in kidney size, may increase in size or **may also end with involution**. **Calcification** is a common feature that develops in adulthood but can happen as early as 3 months after birth.

The histological examination usually reveals **abnormalities in ductal differentiation and minimal corticomedullary differentiation**. The kidney is usually large in size and **resembles a collection of grapes**.

Clinical Presentation and Symptoms of MCDK

At the present time, the disease is detected before birth and as **early as 15 weeks of gestation with ultrasound**. Before the advent of ultrasonography, newborns presented with a flank mass that is mobile, irregular in shape, non-tender and that could transilluminate.

Cases that are not detected on fetal ultrasound can present with **urinary tract infections, hypertension** or **dysfunction with voiding**. They can also be found incidentally on imaging. MCKD is **usually asymptomatic**.

Laboratory Evaluation and Diagnosis of MCDK

Ultrasound is recommended as a preliminary diagnostic study. It shows a random arrangement of cysts of variable size and the **renal pelvis** cannot be identified. An **annual blood pressure measurement** is also recommended. Patients also should have ultrasound every 6 to 12-months until involution of the affected kidney.

Therapy and Treatment of MCDK

Nephrectomy is only performed for symptomatic patients and patients with complications. Those include abdominal or flank pain, urinary tract infections, hypertension and renal cancer.

Prognosis for MCDK

Prognosis is favorable and most cases of unilateral MCKD will have low morbidity and mortality. Morbidity is mostly caused by complications such as **urinary tract infections, hypertension or neoplasia**.

Acquired Cystic Kidney Disease (ACKD)

Definition and Epidemiology of ACKD

ACKD is defined by the development of numerous cysts without prior history of hereditary renal cystic diseases. It usually predates end-stage renal failure and is commonly found incidentally in asymptomatic individuals. It has a significant **association with dialysis** and **renal cell carcinoma**. Other risk factors include male gender and African American ethnicity.

Etiology and Pathophysiology of ACKD

It is thought that **uremia** is the primary cause of ACKD. The association with dialysis is due to the fact that it prolongs the patient's survival. Nonetheless, the exact mechanism remains unknown but is thought to involve ischemia and compensatory growth.

Presentation of ACKD

ACKD is asymptomatic in the early stages. It is present in every type of renal disease that causes a progressive loss of **kidney function**. Kidney involvement is usually **bilateral** and kidneys rarely become palpable.

Diagnosis and Laboratory Evaluation of ACKD

Ultrasonography is the diagnostic modality of choice. It will typically reveal **no increase in renal size**, as well as **normal parenchyma**. On histology, cysts are seen in the cortex, as well as in the medulla with deposition of **oxalate crystals**.

Therapy and Treatment of ACKD

There are no specific treatment recommendations for ACKD. Treatment is directed against loss of kidney function, as well as complications such as **renal cell carcinoma**, **cystic infections**, and bleeding episodes.

Prognosis for ACKD

ACKD may result in significant complications. These include **cystic infections**, **hemorrhage**, **erythrocytosis** and **malignant transformation**.

Simple and Complex Cysts

Background and Epidemiology

Imaging techniques have become preponderant and, as such, asymptomatic cysts are routinely found. These cysts can be classified based on their radiologic characteristics and require different treatment. **Simple renal cysts are far more common** and their incidence increases linearly with age, with a prevalence of more than 30% in elderly individuals (> 70 year old). **Complex cysts are associated with renal cell carcinoma**.

Etiology and Pathophysiology

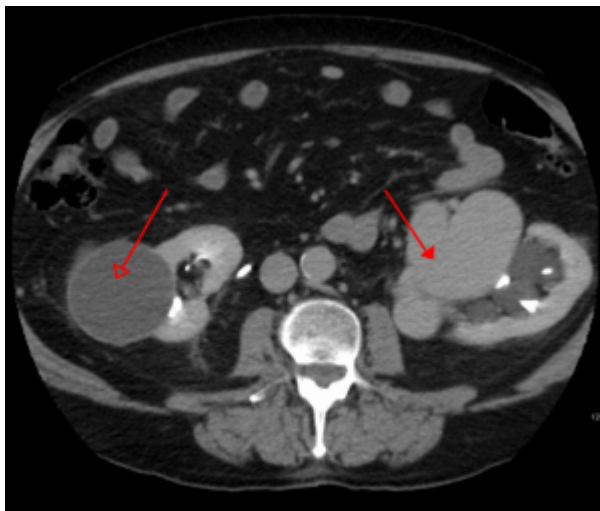
Causes for **simple renal cysts** remain unknown. One popular hypothesis postulates that they start forming from **diverticula** in the collecting tubules and collecting ducts. Simple

cysts can also be present at birth, although occurrence between birth and the age of 20 is very rare. They are typically cortical and extend beyond the parenchyma and are filled with a homogeneous transudate. Studies have shown that they tend to increase very slowly in size.

Complex cysts, on the other hand, have distinguishing characteristics. They tend to show **septae, calcifications, loculation** and **thickening of the walls**. They also show **increased density** after imaging with contrast.

Diagnosis and Laboratory Evaluation

Simple or complex renal cysts are typically found incidentally on imaging, usually with **CT scanning**. A MRI can give a better resolution if further investigation is required to determine the stage of a complex cyst.



[Image](#): "An arterial venous malformation of the left kidney and a simple cyst of the right kidney" by James Heilman, MD. License: [CC BY-SA 3.0](#)

Treatment and Prognosis

Simple cysts usually require no follow-up unless they are symptomatic or enlarging. Complex cysts, on the other hand, may require **follow-up or surgical intervention** due to the risk of renal cell carcinoma.

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