

# Polycystic Kidney Disease

BIRTH DEFECT RESEARCH FOR CHILDREN



## What is Polycystic Kidney Disease?

Polycystic Kidney Disease (PKD) is a genetic disease characterized by the growth of numerous fluid-filled cysts that can result in malformed or enlarged kidneys. These cysts can slowly replace the mass of both kidneys, reducing their function and leading to kidney failure. PKD can also cause cysts in the liver and affect the spleen, the pancreas, the heart, and the blood vessels in the brain.



# Polycystic Kidney Disease



There are two types of PKD. Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common type, accounting for 90% of all PKD cases. Symptoms of the disease usually develop around age 30-40 but can begin as early as childhood. Because of the late development of symptoms, ADPKD is often called “the adult type PKD.” The second type, Autosomal Recessive Polycystic Kidney Disease (ARPKD) is very rare with symptoms usually developing in the womb or during the first few months of life.

How many children have Polycystic Kidney Disease?

ADPKD affects between one in 400 and one in 1,000 people. ARPKD occurs in one in 10,000 births. PKD occurs in all races and affects males and females equally.

How do you know if your child has Polycystic Kidney Disease?

The diagnosis of PKD is usually based on a physical examination, family history, and ultrasound scanning. Difficult cases may require kidney and liver biopsies for diagnosis. Ultrasound scanning of the kidneys of a fetus or newborn can reveal cysts but cannot differentiate between cysts of ADPKD and cysts of ARPKD. ADPKD can go undiagnosed for many years since a person with this disease may have no symptoms. Once cysts have formed, however, ADPKD can be diagnosed based on a combination of kidney cysts, cysts in other organs, and family history. In the future, DNA testing may be able to confirm a diagnosis of ADPKD before the presence of cysts. This technology holds promise for individuals who want to donate kidneys to affected family members. Because ARPKD tends to scar the liver, ultrasound scans of the liver assist in the diagnosis of ARPKD. Severely affected newborns with ARPKD often also have underdeveloped lungs. Children first developing ARPKD symptoms in childhood experience frequent urination, urinary tract infections, and hypertension (high blood pressure). They are also usually smaller than average size.

What causes Polycystic Kidney Disease?

PKD is an inherited disease. In the dominant form (ADPKD) only one parent needs to have the abnormal gene to pass on the disease to their child. In the recessive form (ARPKD) both parents must carry the abnormal gene for the child to get the disease. Genetic counseling is recommended for couples with a family history of PKD.

How can you help a child with Polycystic Kidney Disease?

There is no cure for PKD. Treatments are available to ease some of the symptoms of the disease. Medication can control hypertension and kidney and bladder infections. Proper diet can improve growth of children with ARPKD, and diet and exercise can help control hypertension. Blood tests are used to monitor kidney function. When kidneys fail, patients must have treatment to prevent death. Treatment options include dialysis and kidney transplantation. For children, automated peritoneal dialysis is usually used. In this process, a machine is used to introduce and remove dialysis fluid during the night. Patients receiving transplantation require anti-rejection medication and regular medical follow-up.

What's in the future for a child with Polycystic Kidney Disease?

Individuals with ADPKD may have no symptoms until age 30-40. Symptoms include hypertension, frequent kidney or bladder infections, side or back pain, blood in urine, increase in abdominal size, and headaches. Management of hypertension, prompt treatment of kidney or bladder infections, and healthy lifestyle can prevent or slow the loss of kidney function and prolong a patient's life. Fifty percent of people with ADPKD will have kidney failure by age 60 and 60% by age 70. Prognosis is limited for individuals with ARPKD.

# Polycystic Kidney Disease



The earlier symptoms occur, the more severe the disease. If a child is born with a severe case, he/she can die shortly after birth due to lung insufficiency. Some children die from infections or organ failure (kidney or liver) before or during early adulthood. Children with milder forms of the disease may have no symptoms until adulthood. Because there is no cure for PKD, treatment of the symptoms usually lasts for life. Kidney dialysis and kidney transplantation are options when renal function fails.

Fact Sheet by:

Birth Defect Research Children,  
Inc.

[www.birthdefects.org](http://www.birthdefects.org)

