PKD FOUNDATION
Polycystic Kidney Disease

PKD IN CHILDREN

Polycystic kidney disease (PKD) is a chronic, genetic disease causing uncontrolled growth of fluid-filled cysts in the kidneys, often leading to kidney failure. There is no cure. Currently, there is no FDA-approved treatment for children. The only life-extending options are dialysis and kidney transplant. PKD affects all racial and ethnic groups equally and affects children as well as adults. PKD in children devastates families and dramatically affects quality of life.

There are two types: autosomal dominant PKD and autosomal recessive PKD.

Georgia was born with ADPKD.

Approximately
600,000
people in America and
12.4 million
people in the world have ADPKD.

1 in 25,000
children have ARPKD.

Ava was born with ARPKD.

ADPKD

Autosomal dominant polycystic kidney disease

Fluid-filled cysts develop and enlarge in kidneys, eventually leading to end-stage renal disease in 50 percent of patients by the age of 50.

ARPKD

Autosomal recessive polycystic kidney disease

Kidney cysts develop, the liver becomes fibrotic and prenatal lung development can be delayed.

ABOUT ADPKD

Autosomal dominant polycystic kidney disease (ADPKD) can be diagnosed at a very young age or even before birth.

Diagnostic and/or genetic tests performed during pregnancy should be done in conjunction with medical counseling so the test results can be completely understood. For example, knowing your baby may have an ADPKD gene does not determine the course or severity of the disease. There are two different groups of children with ADPKD — those diagnosed before birth or in their first year of life with large cystic kidneys and those diagnosed after their first year.

GENETICS

When a parent has ADPKD, every conception carries a 50 percent chance of the child inheriting the mutated gene and having ADPKD. In ADPKD, only one mutated gene of the pair is required to pass on the disease. Approximately 10 percent of patients with PKD have no family history of the disease, as ADPKD may develop as a spontaneous (new) mutation.

SCREENING AND DIAGNOSIS

Diagnoses are usually made using radiology — ultrasound, CT scan or MRI.

Characteristics of children diagnosed in first year of life

- Most are diagnosed in-utero with large kidney cysts. Most develop high blood pressure (hypertension) in childhood, which should be monitored and treated by a doctor or pediatric nephrologist.
- Some patients may develop end-stage renal disease by their teenage years.

Characteristics of children diagnosed after first year of life

- Usually, one of the parents is known to be affected with typical ADPKD.
- Some children come to medical attention due to ADPKD related issues like high blood pressure or blood in the urine. In other children, the diagnosis is an "incidental" finding due to widespread use of ultrasounds and improved resolution of the scans to detect smaller-sized cysts.
- Almost all children who are diagnosed after the first year of life will have normal kidney function throughout childhood.

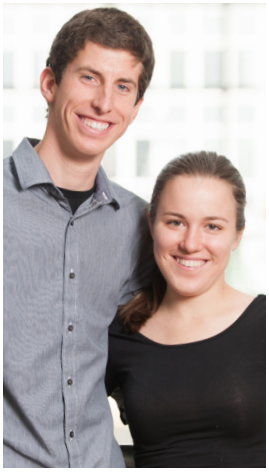
Friends Lena and Brittnee (left) were born with PKD.

SYMPTOMS OF ADPKD

- High blood pressure (hypertension)
- Frequent urinary tract infections
- Back/flank pain
- Mitral valve prolapse
- Kidney stones
- Depression and anxiety (due to stress and emotional impact)
- Enlarged kidneys
- Blood in urine (hematuria)
- Protein in urine (proteinuria)
- Hernia

HEALTH MANAGEMENT

- Have regular appointments with a pediatric nephrologist.
- Maintain a healthy diet: Avoid too much dietary salt and increase water intake.
- Ensure all medications are taken exactly as prescribed.
- Report any and all unusual circumstances to the doctor.



Friends Greg and MaryKatherine (left) were born with ADPKD.

ABOUT ARPKD

Autosomal recessive polycystic kidney disease (ARPKD) is a rare form of PKD, affecting approximately 1 in 25,000 children.

ARPKD can cause death in the first month of life. However, almost 80 percent of ARPKD children survive the newborn period. Previously thought to be a fatal condition, the prognosis for children with ARPKD has improved dramatically. Twenty years ago, only half of the children born with the disease survived to their 10th birthday, but now, more than 90 percent of children who survive the newborn period reach their 20th birthday and 50 percent of these survivors do not yet require dialysis or transplantation.

GENETICS

Each conception carries a 25 percent chance of the child inheriting both mutated copies of the gene. In recessive genetic disorders such as ARPKD, affected children must inherit a copy of the disease gene from both parents. Since the parents each have only one copy of the disease gene, they do not have the disease and are referred to as "carriers." With each conception, there is a 50 percent chance the child will be a carrier and a 25 percent chance both mutated copies of the genes are inherited.

SCREENING AND DIAGNOSIS

KIDNEY Ultrasound, MRI, CT scan
LIVER MR choliangiography

Typically in ARPKD, the kidneys appear to be larger than normal. In some babies, prenatal ultrasound can detect the enlarged kidneys as early as 18 weeks after conception. After birth, the kidneys may be large enough to examine by touch or through an ultrasound. Prenatal genetic testing is possible. These genetic tests involve a direct search of the gene for mutations. For linkage analysis, DNA samples are required from the fetus, a previously affected child, and the parents.

Madeline (above) and Cleo (right) were both born with ARPKD.

SYMPTOMS OF ARPKD

Prenatal

- Diminished amniotic fluid levels during pregnancy
- Enlarged kidneys on fetal ultrasound

Immediately after birth

- Enlarged kidneys due to cysts
- Breathing problems due to poor lung development
- Ventilation frequently required to sustain life
- Enlarged spleen with low red blood cell, white blood cell, and platelet counts
- Congenital hepatic fibrosis (CHF)
- Hypertension
- Growth problems

Beyond newborn period

- Issues with kidney function
- High blood pressure (hypertension)
- Congenital hepatic fibrosis
- Enlarged spleen with low red blood cell, white blood cell, and platelet counts
- Growth problems (for some)

HEALTH MANAGEMENT

Families should work with specialists in several areas, including liver, kidney and nutrition.

PKD FOUNDATION SUPPORT

The PKD Foundation is the only organization in the United States solely dedicated to finding treatments and a cure for polycystic kidney disease.

Since 1982, we have been dedicated to supporting and improving the lives of patients and families impacted by PKD. These efforts are accomplished through promoting research to find treatments and a cure, as well as providing education, advocacy, and awareness on a national level. We provide direct services to local communities nationwide and are the largest private funder of PKD research.

The Foundation supports PKD-relevant scientific and clinical meetings which bring together PKD scientists from around the world to discuss current findings and encourage research collaboration. Additionally, the Foundation has an online resource featuring numerous webinars and videos for families, along with an extensive website.

PKD PARENTS CHAPTER

The PKD Parents Chapter provides support for parents of children with both ARPKD and ADPKD.

This Chapter has served more than 500 families since 2000. Once a family with a child with PKD reaches out to the Foundation, a volunteer from the PKD Parents Chapter contacts them for support, compassion and guidance. Families can be connected with experts and other local families and are supported in many ways.

pkdparents@pkdcure.org

CONNECT

Social media

facebook.com/groups/pkdinchildren

Talk with other parents of children with PKD.

Voices of PKD

Find stories about people affected by PKD.

connect.pkdcure.org/voices-of-pkd

pkdcure.org/pkd-in-children

Since 2006, the PKD Foundation has invested over **$2 million** *in research, support, education, and awareness for ARPKD, ADPKD in children, and congenital hepatic fibrosis (CHF), a disease associated closely with ARPKD.*