POLYCYSTIC KIDNEY DISEASE IN CHILDREN

Polycystic kidney disease (PKD) is one of the most common, life-threatening genetic diseases affecting thousands in America and millions worldwide. There is no treatment or cure for PKD.

In autosomal dominant PKD (ADPKD), fluid-filled cysts develop and enlarge in both kidneys, eventually leading to end-stage renal disease in 50 percent of patients by the age of 50.

In autosomal recessive PKD (ARPKD), kidney cysts develop, the liver becomes fibrotic and prenatal lung development is delayed.

PKD affects children as well as adults. Cystic kidney disease in children devastates families, and dramatically affects quality of life for children who have it. For those who have lost a child, their lives are changed forever.

AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE (ADPKD)

ADPKD can be diagnosed prenatally or at a very young age. Any diagnostic tests done during pregnancy should be done along with medical counseling so the results of the test can be completely understood. The number of cysts a child has affects his/her signs and symptoms. Just as in adults, children with many cysts are more likely to have back, side or stomach pain and are also more likely to have high blood pressure than children with only a few cysts. Almost all children who are diagnosed after the first year of life have perfectly normal kidney function that seems to stay normal throughout childhood. Most children will maintain normal kidney function at least until they are into their mid-20s.

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 PKD gene of the pair is required to pass on the disease.

Diagnosis and Screening

Diagnosis is most often made through radiology, such as an ultrasound, CT scan or MRI.

Characteristics of children diagnosed in first year of life:

- Most of these children have brothers and/or sisters who are also diagnosed in the first year of life.
- Most are diagnosed before birth with large kidneys, but often they do not have actual cysts.
- Most develop high blood pressure in childhood.

Characteristics of children diagnosed after one year of age:

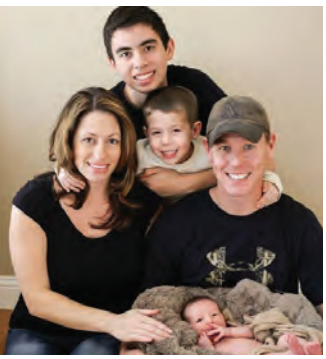
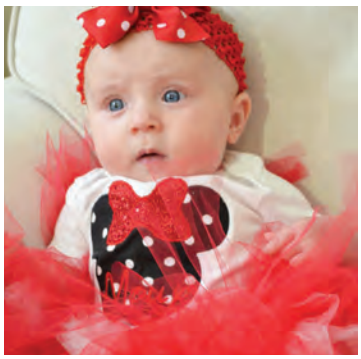
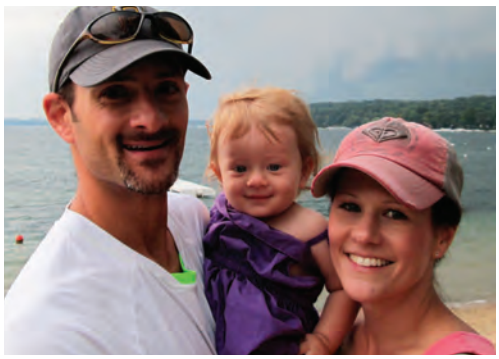
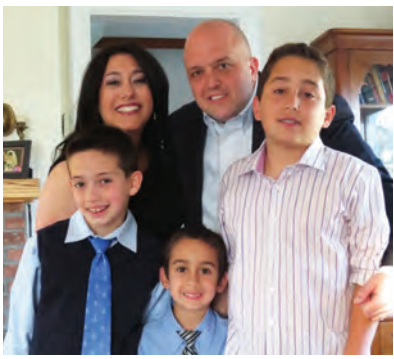
- Just as likely to have an affected father as an affected mother.
- Likely to have cysts, even though their kidneys are not necessarily enlarged.
- Affected with only one cyst (in half the children), or just a few cysts. Whereas, in an adult one cyst alone is not enough to diagnose ADPKD, in children who are part of an ADPKD family, even one cyst means they are more likely to have the disease.

Symptoms of ADPKD

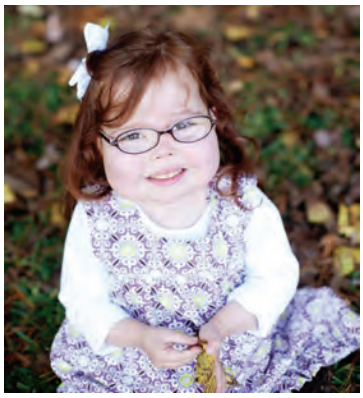
- High blood pressure
- Frequent urinary tract infections
- Blood in urine
- Mitral valve prolapse
- Hernia
- Back/flank pain
- Kidney stones
- Proteinuria (excess protein in urine)
- Enlarged kidneys found during abdominal exam
- Depression and anxiety (due to stress and emotional impact)

Health Management in ADPKD

- Live a healthy life-style, watching diet and including moderate exercise
- Monitor blood pressure; controlling it with medication if it is elevated
- Avoid too much salt, it can increase blood pressure
- Limit caffeine intake

AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE (ARPKD)



ARPKD is a relatively rare form of PKD, affecting approximately 1 in 20,000 children. ARPKD often causes death in the first month of life. For the 70% of ARPKD children who survive the newborn period, approximately one-third will need dialysis or transplantation by age 10. 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 that percentage has increased to 85 percent.

Genetics

Each conception carries a 25 percent chance of the child inheriting both mutated genes and having ARPKD.
In recessive disorders such as ARPKD, the child must inherit a copy of the disease gene from both parents in order to be affected. 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 there will be no mutated genes inherited.

Diagnosis

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 using samples from either chorionic villus sampling or amniocentesis. These genetic tests can either involve a direct search of the gene for mutations or an indirect association using linkage analysis. For linkage analysis, DNA samples are required from the fetus, a previously affected child, and the parents.

Screening

- Ultrasound
- CT scan
- MRI
- MR Cholangiography (x-ray examination of the bile ducts)

Symptoms of ARPKD

Prenatally

- Diminished amniotic fluid levels during pregnancy
- Enlarged kidneys on fetal ultrasound
- Lung immaturity and functioning issues

Immediately after birth

- Enlarged kidneys due to cysts
- Breathing problems due to lack of space because of enlarged kidneys and decreased urine production. Ventilation is frequently required to sustain life.
- Excessive urine production
- Hypertension
- Growth problems
- Congenital hepatic fibrosis
- Enlarged spleen with low red blood cell, white blood cell and platelet counts.

Health Management in ARPKD

- Families should work with specialists in several areas, including liver, kidney and lung specialists, to monitor the child's health.

PKD Foundation Support

The PKD Foundation is the only organization in the U.S. solely dedicated to finding treatments and a cure for PKD to improve the lives of those it affects. We do this through promoting programs of research, education, advocacy, support and awareness on a national level, along with direct services to local communities across the country. Our vision is that one day no one will suffer the full effects of PKD.

The Foundation has invested nearly $2 million since 2006 for research, support, education and awareness for ARPKD, ADPKD in children and congenital hepatic fibrosis (CHF), a disease closely associated with ARPKD.

The Foundation supports PKD-relevant scientific and clinical meetings that bring together PKD scientists from around the world to discuss current findings and encourage research collaboration. In August of 2013, the Foundation supported the International Pediatric Nephrology Association's 2013 World Congress of Nephrology Meeting held in Shanghai, China.

Additionally, the Foundation is an online resource featuring numerous webinars and videos for families, along with an extensive website. Visit **pkdcure.org/learn** for more.

PKD Parents Chapter

The PKD Parents Chapter provides support for parents of children with ARPKD as well as ADPKD.

This Chapter has served more than 500 families since 2000. Once a family with a child living 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. To learn more, email **pkdparents@pkdcure.org**.

Connect

- Email **pkdparents@pkdcure.org.**
- Talk to other parents on the PKD Foundation's Discussion Forums, **pkdcure.org/connect/pkd-connection.**
- Visit **pkdcure.org**.
- Call **1.800.PKD.CURE** (753.2873).
- Follow us on Facebook and Twitter (**@PKDFoundation**).
- Visit Voices of PKD for stories about people affected by PKD: **pkdcure.org/voicesofpkd**.