

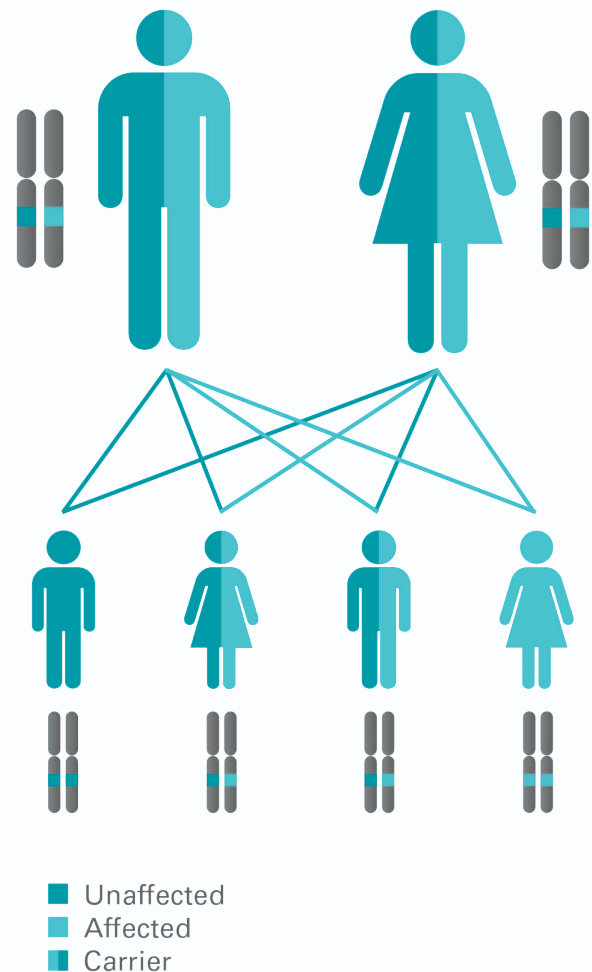
Polycystic Kidney Disease (PKD) affects more than just adults. An estimated 1 in 500 children have the dominant form of PKD, known as autosomal dominant polycystic kidney disease (ADPKD) and another 1 in 20,000 newborns and children inherit a relatively rare form, autosomal recessive polycystic kidney disease (ARPKD).

Although ARPKD isn't as common, it can be very serious with many of the challenges coming in the first month of life. Children who survive the newborn stage have a strong chance of survival overall, although approximately one-third will need dialysis or transplantation by the age of 10.

The following information provides an overview of ARPKD and cystic kidney disease in children. For more in-depth information, including videos and educational resources, visit the ARPKD Learning Center at www.pkdcure.org/arpkd.

ARPKD Genetics

A child inherits ARPKD when both parents have a copy of the disease gene. Since the parents each have only one copy of the disease gene, they do not have the disease and are referred to as "carriers." Two parents who carry the ARPKD gene have a 25 percent chance that each child will inherit the disease.



ARPKD

- ARPKD affects both the kidneys and the liver. In the kidneys, cysts form, causing the kidneys to grow considerably in size and eventually fail. At birth, children with enlarged kidneys and decreased urine production may also experience lung impairment, resulting in critical breathing difficulties. Ventilation is frequently required to sustain life.
- Because of the abnormalities in their kidneys, children with ARPKD often produce excessive amounts of urine during both daytime hours and while sleeping. In addition, many children have difficulty with growth, some of whom require caloric supplements, feeding assistance, and/or growth hormone. As kidney function inevitably declines, medications are often required to do some of the work the kidney is no longer able to keep up with.
- High blood pressure is also very common in children with ARPKD. Current research indicates that untreated hypertension can lead to kidney failure more quickly than if the blood pressure is kept within normal range. Often times medication is required to do so.
- Children with ARPKD also have a liver abnormality called congenital hepatic fibrosis (CHF) that may eventually lead to enlargement of the liver and spleen. CHF can impede the blood flow from the intestine to the liver, causing an increase in pressure and distention of the veins (varices) around the esophagus, stomach and intestine. This is called portal hypertension.
- If the pressure becomes too great, the varices can rupture causing bleeding within the gastro-intestinal cavity. Sometimes portal hypertension leads to splenic enlargement which results in low red blood cells, white blood cells and platelet counts.



Support & Education

The PKD Foundation has an ARPKD Chapter for families who want to connect with others for support, education, advice and more. This chapter was created to serve all families who have children with cystic kidney disease. Led by two passionate ARPKD mothers, the Chapter provides help and hope to families worldwide. To learn more, contact Chapter Coordinators Julia Roberts and Michele Karl at the e-mail address below:

 ARPKDChapter@pkdcure.org

 www.pkdcure.org/arpkd

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Cystic Kidney Disease or ADPKD in Children

It was once thought that people who inherited the gene for ADPKD did not form cysts or have symptoms until well into adult life. We now know that ADPKD can be diagnosed at a very young age and even before birth. Any diagnostic tests done during pregnancy should be done in conjunction with medical counseling so the results of the test can be completely understood. There seems to be two different groups of children with ADPKD – those who are diagnosed before birth or in the first year of life with large kidneys and/or cysts, and those who are diagnosed after the age of 1.

Children who are diagnosed in the first year of life have some special characteristics:

- Most often, the parent with ADPKD is the mother.
- Most of these children have siblings 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, so this should be watched for and treated.

Children who are diagnosed after 1 year of age are:

- 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. While one cyst alone is not enough to diagnose an adult (a few kidney cysts often develop as a natural part of aging), one renal cyst in a child from a PKD family is enough for diagnosis.
- 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.