



### **What is polycystic kidney disease, autosomal recessive?**

Polycystic kidney disease, autosomal recessive (ARPKD) is an inherited disease that appears to be caused by a defect in the protein fibrocystin<sup>1</sup>, which is essential for kidney and liver function.

### **What are the symptoms of ARPKD and what treatment is available?**

Polycystic kidney disease, autosomal recessive (ARPKD) is characterized by multiple cysts in the kidneys leading to symptoms with varying degrees of severity and which present at varying age.

Prenatally diagnosed ARPKD is characterized by<sup>2</sup>:

- Decreased amniotic fluid
- Enlarged kidneys that look brighter than usual on ultrasound
- Underdeveloped lungs resulting in death shortly after birth for about one third of affected newborns

Infantile ARPKD is characterized in part by<sup>3,4</sup>:

- Chronic lung disease
- High blood pressure
- Onset of kidney failure in almost half of individuals by their teens

Childhood and early adulthood ARPKD<sup>3</sup> includes the following characteristics:

- Kidney and liver dysfunction
- Urinary tract infections
- High blood pressure
- Increased risk for internal bleeding
- Infections
- Enlarged spleen

There is no cure for ARPKD, but available treatments include medications for controlling blood pressure and fighting some infections common with this disease. Dialysis or kidney transplantation, and liver transplantation may improve symptoms.<sup>4</sup>

### **How is ARPKD inherited?**

ARPKD is an autosomal recessive disease caused by mutations in the *PKHD1* gene<sup>1</sup>. An individual who inherits one copy of a *PKHD1* gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two mutations in this gene, one from each parent, is expected to be affected with ARPKD.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy. Therefore, it is especially important that the reproductive partner of a carrier be offered testing.

### **Who is at risk for ARPKD?**

ARPKD can occur in individuals of all races and ethnicities.<sup>1,5</sup> The incidence of ARPKD is estimated at 1 in 20,000, and the carrier frequency at about 1/70.<sup>6</sup>

### **What does a positive test result mean?**

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.





### What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

### Where can I get more information?

- Genetics Home Reference: [www.ghr.nlm.nih.gov](http://www.ghr.nlm.nih.gov)
- Genetics and Rare Diseases Information Center (GARD): [www.rarediseases.info.nih.gov](http://www.rarediseases.info.nih.gov)
- ARPKD/CHF Alliance: [www.arpkdchf.org](http://www.arpkdchf.org)
- PKD Foundation: [pkdcure@pkdcure.org](mailto:pkdcure@pkdcure.org)
- PKHD1 database: <http://www.humgen.rwth-aachen.de>

### References

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4. Dell KM and Avner ED. Polycystic kidney disease, autosomal recessive. *GeneReviews.* Available at <http://www.ncbi.nlm.nih.gov/books/NBK1326/>. Accessed: Jan 12, 2012.
5. Bergmann C *et al.* Spectrum of mutations in the gene for autosomal recessive polycystic kidney disease (ARPKD/*PHKD1*). *J Am Soc Nephrol.* 2003; 13: 76-89.
6. Zerres K *et al.* Prenatal diagnosis of autosomal recessive polycystic kidney disease (ARPKD): molecular genetics, clinical experience, and fetal morphology. *Am J Med Genet.* 1998; 76: 137-144.

