

What is autosomal recessive polycystic kidney disease?

Autosomal recessive polycystic kidney disease (ARPKD) is an inherited disease of variable severity and age at onset, characterized by multiple cysts in the kidneys. Individuals with ARPKD have defects in fibrocystin,^{4,7} a protein essential for kidney and liver function.

What are the symptoms of autosomal recessive polycystic kidney disease and what treatment is available?

Symptoms of prenatally diagnosed ARPKD may include:²

- Decreased amount of amniotic fluid
- Enlarged kidneys that look brighter than usual on ultrasound
- Underdeveloped lungs, potentially leading to death shortly after birth

Symptoms of infantile ARPKD may include:^{3,4}

- Chronic lung disease
- High blood pressure

Onset of kidney failure in the teens Symptoms of childhood and early adulthood ARPKD may include:³

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

There is no cure for ARPKD. 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.⁴

How is autosomal recessive polycystic kidney disease inherited?

ARPKD is an autosomal recessive disease caused by mutations in the *PKHD1* gene.¹ 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 *PKHD1* mutations, one from each parent, is expected to be affected with ARPKD.

If both members of a couple are carriers of a mutation in the same gene, the risk of having 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 autosomal recessive polycystic kidney disease?

ARPKD can occur in individuals of all races and ethnicities.^{1,5} The incidence of ARPKD is estimated at 1 in 20,000, and the carrier frequency at about 1 in 70.⁶

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 and Rare Diseases Information Center (GARD): <https://rarediseases.info.nih.gov/gard/8378/autosomal-recessive-polycystic-kidney-disease/resources/1>

ARPKD/CHF Alliance: www.arpkdCHF.org

PKD Foundation: <http://www.pkdcure.org>

References

1. Sharp AM *et al.* Comprehensive genomic analysis of *PKHD1* mutations in ARPKD cohorts. *J Med Genet.* 2005; 42:336-349.
2. Bergmann C *et al.* PKHD1 mutations in autosomal recessive polycystic kidney disease (ARPKD). *Hum Mutat.* 2004; 23: 453-463.
3. Adeva M *et al.* Clinical and molecular characterization defines a broadened spectrum of autosomal recessive polycystic kidney disease (ARPKD). *Medicine.* 2006; 85: 1-21.
4. Sweeney WE and Avner ED. Polycystic kidney disease, autosomal recessive. *GeneReviews.* Available at <http://www.ncbi.nlm.nih.gov/books/NBK1326/>. Accessed: Sept. 22, 2016.
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.
7. PKHD1 gene. Genetics Home Reference. <https://ghr.nlm.nih.gov/gene/pkhd1-conditions>. Published September 20, 2016. Accessed September 28, 2016.