Autosomal Recessive Polycystic Kidney Disease

ARPKD is a rare genetic disorder. It is characterized by kidney, liver, and lung problems as well as urinary tract infections and high blood pressure. A person must have two variants in the PKHD1 gene in order to have this condition.

play+79722d103a, you **do not have** two of the variants we tested.

We tested three variants, but your result could **not be determined** for one of the variants.

The test may not be able to determine a result for every variant tested. This can be caused by random test error, other factors that interfere with the test, or if you have two copies of a tested variant.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial

See Scientific Details

Intended Uses

- Tests for **multiple variants** in the PKHD1 gene.
- To identify carrier status for ARPKD.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** other types of polycystic kidney disease.

Important Ethnicities

- This test does **not** include a large fraction of PKHD1 variants that cause ARPKD in any ethnicity.

We could not determine your result for all variants tested.

ARPKD may still be relevant for you because the variants that cause it are more common in people with **European** ancestry.

You are not a carrier for two of the variants for ARPKD.

We could not determine your result for one of the tested variants.

You still have a chance of being a carrier.

The average person of **European** descent has a **1 in 70 chance** of carrying a variant for ARPKD. Your chances are likely smaller because we ruled out some of the tested variants.

See Scientific Details

About Autosomal Recessive Polycystic Kidney Disease

When symptoms develop

Symptoms typically develop before birth or during infancy.

Typical signs and symptoms

- Kidney disease
- Liver disease
- Respiratory problems
- High blood pressure
- Urinary tract infections

Ethnicities most affected

This condition occurs in people of all ethnicities, but is best studied in people of Finnish, European, Hispanic, Turkish, and Middle Eastern descent.

How it's treated

There is currently no known cure. Treatment focuses on managing the symptoms of kidney, lung, and liver disease, as well as managing blood pressure.

Read more at: Genetics Home Reference GeneReviews

Consider talking to a healthcare professional if you are concerned about your results.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

Print report

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more

Print

Autosomal Recessive Polycystic Kidney Disease

Scientific Details

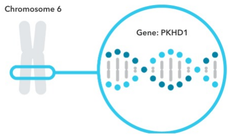
ARPKD is a rare genetic disorder. It is characterized by kidney, liver, and lung problems as well as urinary tract infections and high blood pressure. A person must have two variants in the PKHD1 gene in order to have this condition.

ARPKD is caused by variants in the PKHD1 gene.

PKHD1

The PKHD1 gene contains instructions for making a protein called fibrocystin that is primarily found in the kidneys. Although its exact function is unknown, it is thought to play an important role in the development and function of the kidneys. Certain variants in PKHD1 disrupt its function.

Read more at Genetics Home Reference*



You do not have two of the tested variants. Your result for one of the tested variants could not be determined.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
T36M **Gene:** PKHD1 **Marker:** rs28939383	**Not determined**		Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 4, 5, 6, 8, 10] | ClinVar*
R496X **Gene:** PKHD1 **Marker:** i5012612	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 5, 8, 11] | ClinVar*
D3230fs **Gene:** PKHD1 **Marker:** i5012610	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 5, 6, 7, 8] | ClinVar*

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

We could not determine your result for one or more variants tested. This can be caused by random test error or other factors that interfere with the test. It can also be caused by having two copies of a variant covered by this test.

The average chance of being a carrier for ARPKD is up to 1 in 70 for people with European ancestry. Because you do not have some of the variants, your chance of being a carrier may be less than this.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Autosomal Recessive Polycystic Kidney Disease is indicated for the detection of three variants in the PKHD1 gene. This test is intended to be used to determine carrier status for ARPKD in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include a large fraction of PKHD1 variants that cause ARPKD in any ethnicity.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Finnish	66%	[1]
European	25%	[1]
Hispanic	22%	[1]
Middle Eastern	<1%	[1]
Turkish	<1%	[1]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. ARPKD Mutation Database*
2. Bergmann C et al. (2003). "Spectrum of mutations in the gene for autosomal recessive polycystic kidney disease (ARPKD/PKHD1)." J Am Soc Nephrol. 14(1):76-89.*
3. Bergmann C et al. (2005). "Clinical consequences of PKHD1 mutations in 164 patients with autosomal-recessive polycystic kidney disease (ARPKD)." Kidney Int. 67(3):829-48.*
4. Garcia-Gonzalez MA et al. (2007). "Genetic interaction studies link autosomal dominant and recessive polycystic kidney disease in a common pathway." Hum Mol Genet. 16(16):1940-50.*
5. Gunay-Aygun M et al. (2010). "PKHD1 sequence variations in 78 children and adults with autosomal recessive polycystic kidney disease and congenital hepatic fibrosis." Mol Genet Metab. 99(2):160-73.*
6. Rossetti S et al. (2003). "A complete mutation screen of PKHD1 in autosomal-recessive polycystic kidney disease (ARPKD) pedigrees." Kidney Int. 64(2):391-403.*
7. Sharp AM et al. (2005). "Comprehensive genomic analysis of PKHD1 mutations in ARPKD cohorts." J Med Genet. 42(4):336-49.*
8. Srinivasan BS et al. (2010). "A universal carrier test for the long tail of Mendelian disease." Reprod Biomed Online. 21(4):537-51.*
9. Sweeney WE et al. (2001). "Polycystic Kidney Disease, Autosomal Recessive." [Updated 2016 Sep 15].*
10. Ward CJ et al. (2003). "Cellular and subcellular localization of the ARPKD protein; fibrocystin is expressed on primary cilia." Hum Mol Genet. 12(20):2703-10.*

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
[redacted]	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for one or more of the following genetic markers may see a genotype at these markers: i5012612, rs28939383. This may also update the overall report result for these customers.
[redacted]	Autosomal Recessive Polycystic Kidney Disease report created.