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.

Erin, you **do not have the variants** we tested.

You could still have a variant not covered by this test.

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.

Important Ethnicities

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

You are likely not a carrier.

This result may be less relevant for you because the variants that cause ARPKD are rarely found in people of your ethnicity.

We ruled out the tested variants for ARPKD.

These variants are very rare in all ethnicities.

You still have a chance of being a carrier for ARPKD.

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.

About Autosomal Recessive Polycystic Kidney Disease

When symptoms develop

Symptoms typically develop before birth or during infancy.

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.

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.

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

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

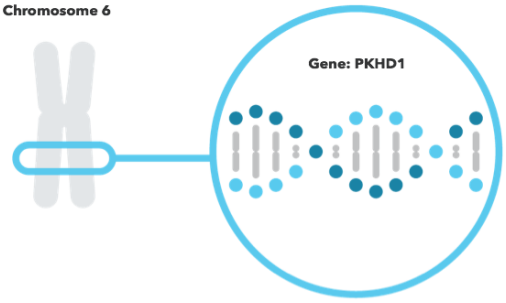
Learn more

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 have no variants detected by this test.

Variants Detected	View All Tested Markers
0	3

Marker Tested	Your Genotype*		Additional Information
T36M Gene: PKHD1 Marker: rs28939383	**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** [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** [2 , 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

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk** .

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of Finnish, European, Hispanic, Middle Eastern, and Turkish descent only.

- For people with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

Finnish	1 in 200	[9]
European	1 in 93	[9]
Hispanic	1 in 89	[9]
Middle Eastern	1 in 70	[9]
Turkish	1 in 70	[9]

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]

Analytical Performance [1]

Analytical accuracy was determined by comparing results from this test with results from sequencing for (Castellani) samples with known variant status. 149 out of 149 genotype results were correct. About 1 in 35,000 samples may receive a **Not Determined** result for one or more variants included in this test. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

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
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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.
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11. Xiong HY et al. (2015). "RNA splicing. The human splicing code reveals new insights into the genetic determinants of disease." Science. 347(6218):1254806.