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.

0 variants detected in the PKHD1 gene

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.

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.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
</table>
| T36M          | G              | Biological explanation  
Typical copy from one of your parents |
| Gene: PKHD1   | Marker: m28939383 | References [2, 4, 5, 6, 9, 10] | ClinVar [1] |
| R496X         | G              | Biological explanation  
Typical copy from one of your parents |
| D2230fs      | T              | Biological explanation  
Typical copy from one of your parents |

*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

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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Frequency</th>
<th>Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finnish</td>
<td>1 in 200</td>
<td>[9]</td>
</tr>
<tr>
<td>European</td>
<td>1 in 93</td>
<td>[9]</td>
</tr>
<tr>
<td>Hispanic</td>
<td>1 in 89</td>
<td>[9]</td>
</tr>
<tr>
<td>Middle Eastern</td>
<td>1 in 70</td>
<td>[9]</td>
</tr>
<tr>
<td>Turkish</td>
<td>1 in 70</td>
<td>[9]</td>
</tr>
</tbody>
</table>
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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finnish</td>
<td>66%</td>
</tr>
<tr>
<td>European</td>
<td>25%</td>
</tr>
<tr>
<td>Hispanic</td>
<td>22%</td>
</tr>
</tbody>
</table>

Reliability was determined by comparing results from this test with results from sequencing for families 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


