Autosomal Recessive Polycystic Kidney Disease

ARPDK is a genetic disorder that's characterized by kidney and long-standing problems as well as other health issues due to high blood pressure. A person who has two variants in the PKD1 gene is more likely to have this condition.

play792270103a, you do not have two of the variants we tested.

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

How To Use This Test

This test does not diagnose any health conditions. Please talk to a healthcare professional if this condition is not in your family, you think you might have these conditions or you have any concerns about your results.

Intended Uses

- Tests to determine genetic risk for the PKD1 gene.
- Tests to identify gene mutations that cause PKD1.

Limitations

- Does not test for all possible variants in the condition.
- Does not report ArPKD for PKD1 alleles.
- Does not assess other types of polycystic kidney disease.

Important Ethicalities

- This test is not advisable to individuals with a family history of PKD1 that involves ArPKD and PKD1.

We could not determine your result for all variants tested.

ARPDK may still be relevant to you because the variants that 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 4 chance of carrying one of the variants. If you are a carrier, there is a 50% chance that your children will have the condition.

About Autosomal Recessive Polycystic Kidney Disease

When symptoms develop

- Symptoms typically develop before birth or during infancy.
- Symptoms include:
  - Kidney disease
  - Liver disease
  - Nephrotic syndrome (kidney disease that affects the liver)
  - Urinary tract infections

When symptoms are absent

- This condition occurs in people of all ethnicities, but is most common in people of European, Hispanic, Japanese, and African-East African ancestry.
- You are at risk if you have a family history of PKD1 and/or PKD2.
- You are at risk if you have symptoms of PKD1 and/or PKD2.

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

If you're a family history of PKD1 and you think you have symptoms, consult with a healthcare professional.

Learn more about the condition and connect with support groups.
APPX2 is a gene located in autosome 7, encoding a transmembrane protein that plays a key role in Alzheimer's disease. The gene is associated with the risk of developing the disease, with its variants potentially influencing the progression and severity of the condition.

**Test Interpretation**

The test determines whether an individual carries at least one copy of the variant APPX2 T226X, which may increase the risk of Alzheimer's disease. It is important to note that the test is not diagnostic and full-genomic analysis is recommended for a comprehensive understanding of an individual's genetic risk.

**Indications for Use**

The test is intended for individuals who have a family history of Alzheimer's disease or are at risk based on genetic or lifestyle factors. It is also recommended for individuals considering genetic counseling or for those interested in understanding their genetic risk.

**Test Performance Summary**

Cystic Fibrosis Carrier Screen & Sickle Cell Trait

- **Sensitivity:** 99% for mutant alleles. This means that the test is highly effective in identifying carriers of the variant.
- **Specificity:** 99% for non-carriers. This indicates a low likelihood of false positives.

**Warranties and Limitations**

- **Warranty:** The test result may not apply to all cases and should be interpreted in the context of a complete medical and genetic evaluation.
- **Limitations:** The test does not account for all genetic factors and may not detect all genetic variants associated with Alzheimer's disease.

**References**

1. HMP study reference.
2.                   

**Change Log**

Your report is successfully updated based on the new information. The following changes have been made to the report:

- **Description:** Updated with the new variant information.

See all references