Pyruvate Kinase Deficiency

Pyruvate kinase (PK) deficiency is a rare genetic disorder in which red blood cells break down too quickly, leading to severe anemia. People with this PK deficiency have two copies of the PKRA gene, one from each parent. In order to have this condition:

**Variant detected**

This test does not diagnose PK deficiency. This result is not expected, please discuss this result with a healthcare professional such as a hematologist who can determine if you have the condition.

How To Use This Test

This test does not diagnose any health conditions. Please discuss the results with a healthcare professional, who can determine if you have any interest in your results.

Intended Uses

- Not used for the ABO blood group in the PKRA gene.
- This test is not for PK deficiency.
- Not for your child’s PK deficiency.
- Not for patients with PK deficiency or with a family history of PK deficiency, patients may be at risk of developing symptoms of PK deficiency.

Limitations

- Not used for the ABO blood group in the PKRA gene.
- This test is not for PK deficiency.
- Not for your child’s PK deficiency.

Important Considerations

- This test does not include the majority of PKRA variants that cause PK deficiency in any condition.

You are at risk of developing symptoms of PK deficiency.

Your result may be relevant if you are considering having children.

Talk to a healthcare professional.

People with your result are at risk of developing symptoms of PK deficiency. As healthcare professionals, we are committed to helping you understand and make informed decisions about your health. We do not provide medical treatment or recommendations about healthcare. If you have questions about your results, see Frequently Asked Questions for more information.

About Pyruvate Kinase Deficiency

Also known as: PK deficiency, PK deficiency

When symptoms develop

Symptoms can develop anytime from birth to adulthood. The severity of the PK deficiency can vary from mild to severe. Symptoms may worsen with age.

Signs and symptoms

- Swelling or pain
- Fatigue and difficulty exercising
- Joint dislocation of the hip and arm
- Encephalitis, encephalopathy, and difficulty concentrating
- Encephalitis
- Eye issues
- Hearing loss

Ethnicity most affected

PK deficiency occurs in many people of many ethnicities. It is most often associated with the African descent, but the condition PK deficiency is more common in the Caucasian and African populations.

How it tested

There is currently no cure. Treatment depends on the severity of the symptoms and can include: blood transfusions, medications to increase exercise in the brain and muscles, and surgery to repair the eye and tissues.

Support groups and resources

Pyruvate kinase deficiency is a genetic disorder that can be managed through proper medical care. It is important to talk to a healthcare professional if you are concerned about your results.

Support groups and resources

- [Genetic and Rare Disease Information Center](https://rarediseases.info.nih.gov)

Read more at: [MedHelp](https://www.medhelp.org)

It is important to talk to a healthcare professional if you are concerned about your results.

Support groups and resources

- [Genetic and Rare Disease Information Center](https://rarediseases.info.nih.gov)

Your result may be relevant if you are considering having children.
Pyruvate Kinase Deficiency

Pyruvate kinase deficiency (PKD) is a rare, inherited disorder that impairs the body's ability to convert glucose into energy. It is caused by mutations in the PKLR gene, located on chromosome 16. These mutations interfere with the production of functional pyruvate kinase, an enzyme that plays a crucial role in the glycolytic pathway.

**Indications for Use**

- The diagnosis of PKD is typically made in patients with symptoms such as weakness, fatigue, and exercise intolerance.
- PKD can be suspected in individuals with a family history of the disorder.
- Blood tests and genetic testing are used to confirm the diagnosis.

**Risks and Limitations**

- PKD is a rare disorder, and genetic testing may not be available in all regions.
- The test may not detect all mutations associated with PKD.
- False-negative results can occur in some cases.

**References**


**Change Log**

- March 17, 2021: Initial release of the PKD test information.
Frequently Asked Questions

Pyrurate Kinase (PK) deficiency is a rare genetic disorder in which red blood cells break down too quickly, leading to chronic anemia. A person must have two variants in the PKLR gene, or two copies of a variant, in order to have this condition.

Overview  Scientific Details  Frequently Asked Questions

Pyrurate Kinase Deficiency

What does this test do?

This test looks for the R484W variant in the PKLR gene linked to PK deficiency.

People with one PKLR variant can pass the variant on to their children.

People with two PKLR variants or two copies of a variant are at risk of developing symptoms of PK deficiency, and they will pass a variant on to each of their children.

This test does not include all possible genetic variants linked to PK deficiency.

Is this answer helpful?  Yes  No

What does this test not do?

This test does not diagnose PK deficiency. Only a healthcare professional can do that.

This test does not include all possible variants in the PKLR gene linked to PK deficiency. It does not include the majority of variants linked to PK deficiency in any ethnicity.

Is this answer helpful?  Yes  No

My report says two copies of a variant were detected. What does this mean?

This means you have two copies of the genetic variant we tested. You inherited one variant from each of your parents. People with this result are at risk of developing symptoms of PK deficiency. It's important to talk with a healthcare professional if you are concerned about your result.

This result also means you will pass a variant on to each of your children.

Is this answer helpful?  Yes  No

What does this test do at risk of developing symptoms of PK deficiency mean?

Symptoms of PK deficiency include chronic anemia, extreme fatigue and difficulty exercising, palmar (yellowing of the skin and eyes), cognitive difficulties such as difficulty concentrating, an enlarged spleen, iron overload, and gallstones.

People with your genetic result are at risk of developing these symptoms. Symptoms can vary widely from person to person and can develop anytime from before birth to adulthood. Symptoms may worsen with age.

A healthcare professional such as a hematologist (a doctor who specializes in conditions that affect the blood) can answer any questions you may have about your results.

Is this answer helpful?  Yes  No

My report says I have two copies of a variant linked to PK deficiency. What are some things I could do?

Based on your genetic result, you are at risk of developing symptoms of PK deficiency. It is important to talk to a healthcare professional if you are concerned about your result.

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

You may also want to share your results with other family members. Because you share DNA with your relatives, your genetic result could also be relevant for them.

Is this answer helpful?  Yes  No

How could my result affect my children?

Because you have two copies of a variant, you will pass a variant on to each of your children. If your partner is a carrier for PK deficiency, each of your children has a 50% chance of having this condition.

A genetic counselor can help you and your partner understand if additional testing might be appropriate. Learn more about genetic counseling.

Is this answer helpful?  Yes  No

Have more questions? Check out our Customer Care Help Center.

Development of the Pyruvate Kinase Deficiency report was supported in part by Aiguo Pharmaceuticals. 23andMe retains sole responsibility for the final report content.