Pyruvate Kinase Deficiency

Pyruvate kinase (PK) deficiency is a rare genetic disorder in which red blood cells break down too quickly, leading to chronic anemia. A person with this condition has mutations in the PKL2 gene, or two copies of a variant in order to have this condition.

Jamie, we could not determine if you have the variant we tested.

This test is intended to detect a variant in the PKL2 gene but your result could not be determined.

How To Use This Test

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

Return the Center Return Where
See Scientific Details

Intended Uses

- To test for the PK deficiency in the PKL2 gene.
- To identify at-risk persons for PK deficiency.
- To inform individuals with two copies of the variant that they may be at risk of developing symptoms of PK deficiency.

Limitations

- This test is not suitable for all possible variants for the condition. More than 300 known PK deficiency genes have been linked to PK deficiency, and this report only includes one of those tests.

Important Ethnicities

- This test does not include the majority of PKL2 variants that cause PK deficiency in any ethnicity.

We could not determine your carrier status.

Your test result could not be determined.

About Pyruvate Kinase Deficiency

Also known as: PK Deficiency, PKD

If you have symptoms:

- Chronic fatigue
- weekend
- Iron deficiency
- Fatigue

If you have symptoms:

- Frequent symptoms
- Fatigue
- Iron deficiency
- Fatigue

If you are concerned about your results:

- If you have symptoms, consult with a healthcare professional.
- If you have a family history of this condition or you have symptoms, consult with a healthcare professional.
- Share your results with a healthcare professional.
- Learn more about this condition and connect with support groups.

Development of the Pyruvate Kinase Deficiency report was supported by AgroPharmaceuticals. 23andMe retains sole responsibility for the final report content.
Pyruvate Kinase Deficiency

Pyruvate Kinase (PK) deficiency is an inherited autosomal recessive disorder that results from a deficiency in PK activity due to inheritable mutations in the PKLR gene. PK deficiency is associated with a number of clinical presentations, including anemia and muscle weakness.

Your result for this test could not be determined.

| Gene Name | PKLR | Condition | Pyruvate Kinase Deficiency

**Test Interpretation**

You have returned "Not determined" results. This indicates that the genetic test did not provide a definitive result for PK deficiency. This could be due to a variety of reasons, including limited test sensitivity or specific genetic mutations.

**Test Details**

**Indications for Use**

- To diagnose Pyruvate Kinase Deficiency
- To screen for PK deficiency in at-risk populations

**Warnings and Limitations**

- The test may not detect all mutations associated with PK deficiency.
- Results should be interpreted in the context of clinical findings.

**References**


**Change Log**

- Title: Pyruvate Kinase Deficiency
- Date: March 12, 2007

The test for Pyruvate Kinase Deficiency is performed by a genetic laboratory. For more information, please consult your healthcare provider.