Pyruvate Kinase Deficiency

Pyruvate kinase (PK) deficiency is a genetic disorder in which the body’s ability to deliver energy to cells is impaired. In PK deficiency, the body has a low level of an enzyme called pyruvate kinase, which plays a crucial role in the process of energy production. If you have PK deficiency, your body may not produce enough energy to function properly, leading to various symptoms and conditions.

How to Use This Test

This test does not diagnose any health conditions. Please consult with a healthcare professional if the condition worsens or if you have any concerns about your health.

Intended Use

• To test for the PK variant in the PKU gene.
• To identify patients with PKU deficiency.
• To determine if a family member has a PKU-related condition.

Limitations

• This test does not identify all possible causes for the condition. Since more than 100 variants in the PKU gene have been linked to PKU deficiency, this report only includes one of these variants.

Important Ethics

• This test does not include the reporting of PKU carriers that may cause PKU deficiency in any offspring.

You are a carrier.

You could pass the variant on to your children.

Your results may be relevant for you if you’re thinking about starting a family.

If you and your partner are both carriers, each child may have a 25% chance of having this condition. Your relatives may be at risk for carrying a PKU-related condition.

We detected one variant for PKU deficiency.

People with only one variant are not expected to have PKU deficiency.

About Pyruvate Kinase Deficiency

This variant is PKU.EV6314, also known as PKU.EV6314.

When symptoms develop

Symptoms can develop from before birth to adulthood, and can vary from mild to severe. Symptoms may appear after age 4.

Optical signs and symptoms

• Vision problems
• Seizures
• Difficulty breathing
• Developmental delays

Cognitive difficulties such as difficulty concentrating

Epilepsy

Fatigability

GBS

Mild defects

PKU is more common in women than in men. There is currently no cure for PKU. Treatment depends on the severity of the symptoms and includes diet modification, frequent blood transfusions, and periods of hospitalization. In severe cases, a lifelong diet may be required. The condition may be treatable with certain medications, but no cure is currently available.

Important ethics

• This test does not include the reporting of PKU carriers that may cause PKU deficiency in any offspring.

Consider telling a healthcare professional if you are thinking about having children.

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

You can learn more about PKU deficiency and how to prevent its transmission to your children by consulting with a healthcare professional.

Learn more about this condition and connect with support groups.

Development of the Pyruvate Kinase Deficiency report was supported by Agios Pharmaceuticals. LuminiteWARNING: This report is for informational purposes only and should not be used as the sole basis for any action.
PK deficiency is caused by variants in the PKLR gene.

You have one variant detected by this test.

**Gene:** PKLR

**Variant:** c.405dupG

**Protein consequence:** Frameshift, possibly truncating

**Functional consequence:** Polypeptide chain slightly unstable

**Phenotype:** None observed

**Clinical significance:** Not significant

**Reference:** 3, 5, 7, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21

**Mendelian: Inheritance**

- PK deficiency is inherited in an autosomal recessive manner.

**Text Details**

**Indications for Use**

- To evaluate PK deficiency in cases presenting with clinical symptoms suggestive of PK deficiency and a negative family history.

**Test Forensic Summary**

**Cancer Screening:** Not indicated.

**Comments:**

- The sensitivity of this test is high, with a specificity of 100%.

**References**