TEST

Pyruvate kinase blood test

A blood test that measures pyruvate kinase enzyme activity in red blood cells

WHAT IT MEASURES
Pyruvate kinase enzyme activity in red blood cells

SAMPLE
Blood from a vein

FASTING NEEDED?
No

HOW LONG IT TAKES
About 5 to 10 minutes

RESULTS IN
Several days to two weeks; varies by laboratory

USED FOR
Diagnosing suspected inherited pyruvate kinase deficiency

MAIN LIMITATION
Transfused or young red cells can mask low activity

Overview

A pyruvate kinase blood test measures the activity of an enzyme called pyruvate kinase inside your red blood cells. Enzymes are proteins that help chemical reactions happen.

Red blood cells need pyruvate kinase to make energy. If the enzyme does not work properly, the cells may break down earlier than expected. This breakdown is called hemolysis and can cause hemolytic anemia.

The test is mainly used when a clinician suspects inherited pyruvate kinase deficiency. This rare genetic condition is linked to changes in the `PKLR` gene. The blood test is not the same as measuring pyruvate in the blood, and it is not a general screening test for anemia.

A low result can support the diagnosis, but the enzyme test is often considered alongside other blood tests and genetic testing. Several factors can make the activity appear normal even when a deficiency is present.

Why the test is ordered

The test may be ordered during an investigation of unexplained hemolytic anemia. It is generally considered after more common causes have been assessed.

Reasons for testing may include:

The test may also help distinguish pyruvate kinase deficiency from other inherited red-cell conditions. It does not identify the severity or likely course of the condition on its own.

- Anemia with evidence that red blood cells are breaking down early
- Unexplained jaundice, meaning yellowing of the skin or eyes
- A high reticulocyte count, showing increased production of young red cells
- Hemolytic anemia beginning in infancy or childhood
- A family history of pyruvate kinase deficiency
- An unclear result from genetic or other red-cell enzyme testing

Who should have the test

This is a specialist test rather than part of a routine blood count. It may be appropriate if your medical history and other laboratory results suggest ongoing red-cell breakdown without a clear explanation.

Testing can be done in children or adults. Family members may sometimes be offered testing after a genetic change associated with pyruvate kinase deficiency has been identified in a relative. In that situation, genetic testing may be more informative than measuring enzyme activity alone.

The test is not normally used to screen people without a relevant personal or family history. A clinician or blood-disorder specialist can decide whether enzyme testing, genetic testing, or both would answer the clinical question.

How to prepare

You do not need to fast. Unless you have been given separate instructions for another test, you can eat and drink normally.

Tell the person arranging the test if you have recently had a blood transfusion. Donated red blood cells may have normal pyruvate kinase activity and can hide a low result in your own cells. The timing of testing may need to be adjusted, or genetic testing may be considered instead.

Also mention:

Do not stop prescribed medicine unless the clinician who prescribed it tells you to.

- Recent major bleeding or a hemolytic episode
- Recent removal of the spleen
- Current medicines, including treatment for pyruvate kinase deficiency
- Any difficulty with previous blood draws

What happens during the test

A healthcare professional places a small needle into a vein, usually in your arm. Blood is collected into a tube containing a substance that prevents clotting.

The collection usually takes a few minutes. The sample must then be handled and transported according to the testing laboratory's instructions because enzyme activity can change if storage conditions are unsuitable.

Pyruvate kinase testing is often performed at a specialist laboratory rather than at the clinic where your blood is collected. This is why results may take longer than a routine blood count.

Understanding your results

Results are usually reported as pyruvate kinase activity relative to the amount of hemoglobin in the sample. Hemoglobin is the oxygen-carrying protein in red blood cells.

There is no single reference range that applies to every laboratory. Methods and units differ. Ranges and interpretation can also vary with the laboratory, age, sex, pregnancy status, medicines, and the wider clinical context.

A low result may support pyruvate kinase deficiency, particularly when there is other evidence of long-term hemolysis. It does not prove that an inherited `PKLR` gene change is responsible. The result may need confirmation with repeat enzyme testing or genetic testing.

A normal result does not always rule out deficiency. Recent transfusion, large numbers of young red cells, or contamination by other blood cells can raise the measured activity.

A high result usually does not mean that you have a pyruvate kinase overactivity disorder. It may reflect a high proportion of young red cells or the way the sample was prepared.

Your result should therefore be compared with your blood count, reticulocyte count, bilirubin, signs of hemolysis, transfusion history, and any genetic findings.

What affects the results

This test is especially sensitive to the mixture of cells in the sample.

A recent blood transfusion is one of the most important influences. Normal donor red cells can increase the measured activity and mask pyruvate kinase deficiency. Tell the testing team the date of your most recent transfusion, even if it occurred at another hospital.

Reticulocytes are young red blood cells. They normally contain more pyruvate kinase than older cells. Hemolysis, recent bleeding, or recovery from anemia can increase the reticulocyte count and make the overall enzyme activity look higher than expected.

White blood cells and platelets also contain pyruvate kinase. If they are not adequately removed during sample preparation, they can produce a falsely reassuring result.

Other influences include:

Normal eating, coffee, and the time of day are not known to be major influences on this enzyme measurement.

- Delayed transport or incorrect sample temperature
- A clotted, damaged, or poorly stored sample
- Recent spleen removal, which can alter the mix of circulating red cells
- Medicines that activate or affect pyruvate kinase
- Differences between laboratory testing methods

Accuracy and limitations

The test can correctly identify markedly reduced enzyme activity when the sample is collected and processed under suitable conditions. However, there is no single published accuracy figure that applies to every laboratory method or clinical situation.

It can miss pyruvate kinase deficiency by giving a normal or near-normal result. This is most likely after a transfusion, when many young red cells are present, or when white cells and platelets remain in the sample.

It can also report activity as lower than it truly is if the specimen deteriorates during storage or transport. A low result alone cannot establish that the cause is an inherited `PKLR` gene change.

Genetic testing is often the better test after a recent transfusion or when enzyme results are unclear. It looks for disease-causing changes in `PKLR`. However, some genetic tests may not

detect every type of change, such as certain large deletions or changes outside the regions routinely examined.

For these reasons, enzyme and genetic results may be used together rather than treated as competing answers.

Risks and complications

The main risks are brief discomfort, a small bruise, or feeling faint during or after the blood draw. Serious complications from routine blood collection are uncommon.

What happens next

Your result will usually be reviewed with the other tests that led to pyruvate kinase testing. If activity is clearly low, the next step may be repeat enzyme testing, `PKLR` genetic testing, or referral to a hematologist, who is a doctor specializing in blood conditions.

If the result is normal but suspicion remains, the clinician may check whether transfusion, reticulocytosis, or sample handling affected it. Testing may be repeated at a more suitable time, or genetic testing may be used.

These are routine follow-up steps and do not usually require emergency care. Seek urgent medical assessment if you develop severe shortness of breath, chest pain, fainting, confusion, or rapidly worsening weakness. These symptoms can have many causes and should not be interpreted from the test result alone.

Common questions

Can I drink coffee before the test?

Yes. Coffee does not usually affect red-cell pyruvate kinase activity. Follow any separate fasting instructions if other tests are being collected at the same appointment.

Does the time of day matter?

No important daily pattern is recognized for this test. The laboratory's transport schedule may matter more because the sample must be processed correctly.

Can I exercise beforehand?

Ordinary activity is unlikely to change the result substantially. If strenuous exercise makes you feel unwell or dizzy, avoid it before the blood draw and tell the person collecting the sample.

How long after a transfusion should I wait?

There is no single waiting period suitable for everyone. It is influenced by the amount and timing of transfused blood and the purpose of testing. Give the clinician and laboratory the exact transfusion date so they can choose the appropriate timing or consider genetic testing.

Can this test tell how severe the condition is?

Not reliably. The measured activity does not consistently predict symptoms, transfusion needs, or future complications. Severity is assessed using your history, examination, blood results, and treatment needs.

Will I also need a genetic test?

Possibly. Genetic testing can confirm relevant `PKLR` changes and is particularly useful after transfusion or when enzyme activity is borderline. Enzyme testing can still provide useful evidence because genetic testing does not detect every possible change.

Sources

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