



PK DEFICIENCY

DIAGNOSTIC TESTING PROGRAM

Diagnostic testing for pyruvate kinase (PK) deficiency is now available from participating third-party laboratories—**at no cost to the patient.**

This program is designed to make testing easier for the patient and healthcare provider by lowering barriers such as:

- Unfamiliarity with labs performing diagnostic testing for PK deficiency
- Lack of local geographic access to testing
- Issues relating to cost of testing

Is PK Deficiency Behind Your Patient's Hemolytic Anemia?
Test to Know.

Diagnostic Testing for PK Deficiency

Now Available at No Cost to the Patient*

About the PK Deficiency Diagnostic Testing Program

Agios is sponsoring no-charge third-party diagnostic testing for pyruvate kinase (PK) deficiency in individuals who may have hemolytic anemia of an unknown cause, when certain criteria are met as requested by a healthcare professional. This program was created to lower barriers to diagnostic testing, helping people and their healthcare providers make informed decisions about their health.

Finding a Participating Laboratory

Agios provides the *Diagnosing PK Deficiency* information sheet, which lists participating labs, online at www.KnowPKDeficiency.com, or by sending a request for this information to knowpkdeficiency@agios.com.

How the Program Works

- The Test Request Form (TRF) for each participating institution contains laboratory-specific specimen requirements and shipping information
- Please refer to the respective test request and specimen handling forms for each participating institution to place an order

Privacy and the Program

While Agios provides financial support for this program, all tests and services are performed by the selected third party. Agios receives contact information for healthcare professionals who submit tests under this program, as well as limited de-identified aggregate patient data. Using the TRF, physicians can elect to share contacts in order to receive information regarding upcoming clinical trials and/or general information about PK deficiency.

PK deficiency could be the underlying cause behind your patient's hemolytic anemia. **Test to confirm or rule out PK deficiency.**

*The cost of testing is covered by Agios when performed by laboratories participating in this program and when program-specific TRFs are used.

Questions and Answers

About the Diagnostic Testing Program

When should a diagnosis of PK deficiency be considered?

PK deficiency should be considered in patients with hemolysis but who lack evidence of an acquired autoimmune disorder. In neonates, consider PK deficiency in cases of persistent hyperbilirubinemia and/or anemia without clear causative factors (even if no reticulocytosis is present).^{1,2}

How is a diagnosis of PK deficiency made?

Biochemical testing is available to diagnose PK deficiency based on enzyme activity and is considered the standard. However, PK enzymatic activity could incorrectly appear normal or high for many reasons, including post-transfusion contamination of patient cells (mutant PK) with healthy donor cells (normal PK) or failure to remove leukocytes from the sample.^{1,3}

Molecular testing, such as *PK-LR* analysis, can help confirm diagnosis in cases where a patient is chronically transfused, has normal or decreased PK enzyme activity in the presence of elevated activity of other age-dependent red cell enzymes, or has low PK enzyme activity but no family history of PK deficiency.^{1,3}

For decisions regarding which test is appropriate for your patient, we encourage you to consult with the laboratories where you are sending the sample—listed in the HCP section of www.KnowPKDeficiency.com.

How do I obtain free testing through this diagnostic support program?

Testing at no cost to the patient is only available through specific laboratories participating in this collaboration and when specific Test Request Forms (TRFs) are used. Participating labs and TRFs are found online at www.KnowPKDeficiency.com/testing or by sending a request for information to knowpkdeficiency@agios.com.



Our Commitment to Researching **PK Deficiency**, an Under-Recognized Rare Disease

Agios is committed to better understanding PK deficiency and the burden experienced by people living with this disease. As part of our broader disease awareness efforts, Agios has created resources to support diagnostic awareness, including information regarding signs and clinical presentation in suspected patients, information on diagnostic methods, and laboratory listings in the United States and abroad. And, in an effort to lower barriers to testing, Agios has entered into collaborations with laboratories to cover the cost of the testing and the reporting of test results.

Learn more at **www.KnowPKDeficiency.com/testing**

References:

1. Grace RF, Zanella A, Neufeld EJ, et al. Erythrocyte pyruvate kinase deficiency: 2015 status report. *Am J Hematol*. 2015;90(9):825-830.
2. Pissard S, de Montalembert M, Bachir D, et al. Pyruvate kinase (PK) deficiency in newborns: the pitfalls of diagnosis. *J Pediatr*. 2007;150(4):443-445.
3. Gallagher PG, Glader B. Diagnosis of pyruvate kinase deficiency. *Pediatr Blood Cancer*. 2016;63(5):771-772.

