

# PK Deficiency Diagnostic Support Program

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

This program is designed to make testing easier by lowering barriers such as:

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

**Do You Have a Patient With PK Deficiency?  
Test to Know.**

*A program sponsored by*



# DIAGNOSTIC TESTING FOR PK DEFICIENCY

## About the PK Deficiency Diagnostic Support 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 to help patients and their healthcare providers make informed decisions about their health.

## How the Program Works

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

## Finding a Participating Laboratory

Participating labs are listed on the Diagnosing PK Deficiency information sheet, available from Agios, online at [www.knowpkdeficiency.com](http://www.knowpkdeficiency.com), or by sending a request for information to [knowpkdeficiency@agios.com](mailto:knowpkdeficiency@agios.com).

## 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 opt in to share their contact information to receive information regarding upcoming clinical trials and/or general information about PK deficiency.



**If you have a patient with hemolytic anemia of unknown cause, it could be PK deficiency. Test to know.**

# Now Available at No Cost to the Patient\*

## Questions & Answers About the Diagnostic Support Program

### ▶ **When should PK deficiency be considered?**

*PK deficiency should be considered in patients with hemolysis but who lack evidence of an acquired autoimmune disorder.<sup>1,2</sup> In neonates, consider PK deficiency in cases of persistent hyperbilirubinemia and/or anemia without clear causative factors (even if no reticulocytosis is present).<sup>1,3</sup>*

### ▶ **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 to diagnose PK deficiency.<sup>1,4</sup> 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.*

*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.<sup>1,4</sup>*

*For decisions regarding which test is appropriate for your patient, consult with the laboratories listed on the Diagnosing PK Deficiency page of the HCP section at [knowpkdeficiency.com](http://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 [knowpkdeficiency.com](http://knowpkdeficiency.com) or by sending a request for information to [knowpkdeficiency@agios.com](mailto:knowpkdeficiency@agios.com).*

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

## Our Commitment in PK Deficiency

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 on diagnostic methods and laboratory listings in the United States and abroad. In an effort to lower barriers to testing, Agios has entered into collaborations with specific laboratories to cover the cost of the test and reporting test results.

**Learn more at [www.knowPKdeficiency.com](http://www.knowPKdeficiency.com)**

1. Grace RF, et al. Erythrocyte pyruvate kinase deficiency: 2015 status report. *Am J Hematol.* 2015;90(9):825-30.
2. Hirono A, Kanno H, Miwa S, and Beutler E. Chapter 182 Pyruvate Kinase Deficiency and Other Enzymopathies of the Erythrocyte. New York, NY: McGraw Hill. 2014.
3. Pissard S et al. Pyruvate kinase (PK) deficiency in newborns: the pitfalls of diagnosis. *J Pediatr.* 2007;150(4):443-5.
4. Gallagher PG and Glader B. Diagnosis of pyruvate kinase deficiency. *Pediatr Blood Cancer.* 2016 May;63(5):771-2.

