

# ILLUMINATING

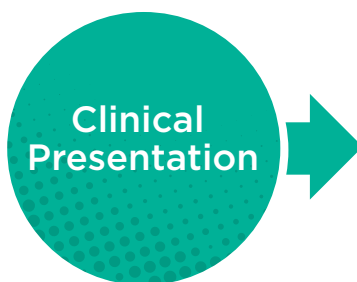
## AN UNDERLYING CAUSE OF HEMOLYTIC ANEMIA

### Diagnosing PK Deficiency

PK deficiency is a lifelong, chronic disease characterized by hereditary hemolytic anemia, which results from a deficiency of the enzyme pyruvate kinase (PK).

Patient presentation is highly variable, ranging from mild to life-threatening, with severe debilitating co-morbidities.<sup>1,2</sup>

The disease may be underrecognized,<sup>3</sup> particularly in adults and patients on the milder end of the spectrum of disease severity.<sup>2</sup>



Anemia  
Dyspnea  
Exercise intolerance  
Abdominal pain  
Iron overload  
Fatigue/weakness  
Jaundice  
Splenomegaly  
Gallstones



**Typically Decreased:**  
Hemoglobin/hematocrit,  
pyruvate kinase (PK)  
activity, haptoglobin

**Typically Elevated:**  
Reticulocytes, platelets,  
bilirubin, MCV, ferritin



Paroxysmal nocturnal  
hemoglobinuria  
Glucose-6-phosphate  
dehydrogenase  
deficiency  
Hereditary spherocytosis  
Autoimmune  
hemolytic anemia  
Beta thalassemias  
Hereditary elliptocytosis

**PK Deficiency**

#### DIAGNOSTIC TESTS

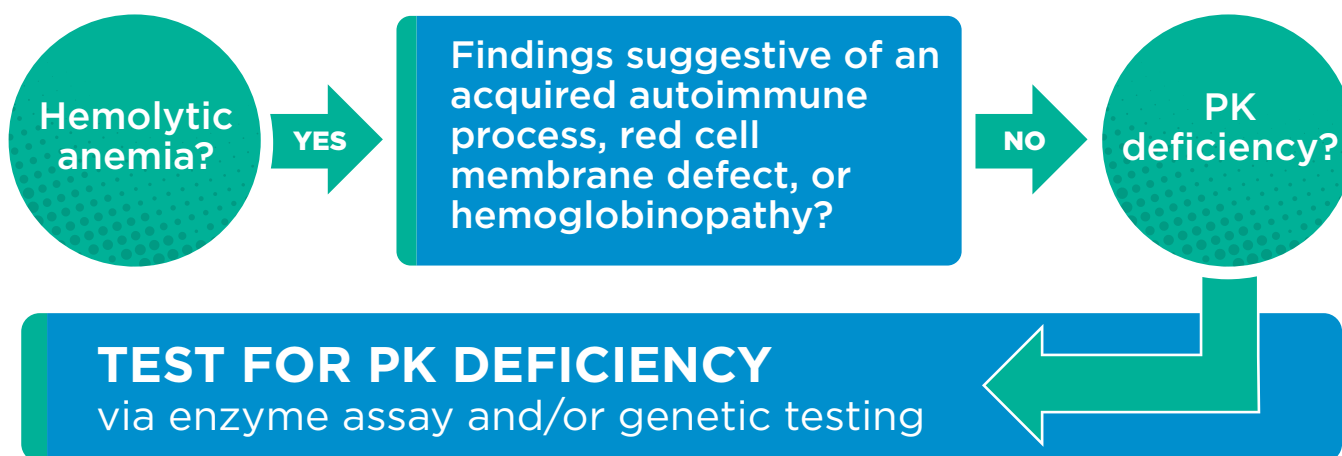
**1st ENZYME ASSAY FOR  
PYRUVATE KINASE  
ACTIVITY**

Enzyme assay is the gold standard for diagnostic testing of PK deficiency.<sup>3</sup>

**2nd MOLECULAR PK-LR  
ANALYSIS**

Genetic testing may be conducted to confirm equivocal cases.<sup>3</sup>

**TEST FOR PK DEFICIENCY**



## Diagnostic Testing for PK Deficiency

Laboratory	Contact Email and Phone	Type of Test
ARUP Laboratories, General Laboratory (Salt Lake City, UT)	Cynthia Gin, BS, MT (ASCP) ginca@aruplab.com 800-242-2787	Enzyme assay
Mayo Clinic, Metabolic Hematology Laboratory (Rochester, MN)	Lea Koon, MS / Michelle Kluge, MS, CGC rstgchemepath@mayo.edu 800-533-1710	Enzyme assay
Cincinnati Children's Hospital Medical Center, Molecular Genetics Laboratory (Cincinnati, OH)	Haley Keller / Chinmayee Nagaraj / Elizabeth Ulm / Emily Wakefield haley.keller@cchmc.org 513-636-4474	Enzyme assay  Genetic Testing (PKLR sequencing)
PreventionGenetics, Clinical DNA Testing and DNA Banking (Marshfield, WI)	Guoli Sun, MD, Ph, FACMG / Angela Gruber, PhD / Bruce Krawisz, MD clinicaltesting@preventiongenetics.com 715-387-0484	Enzyme assay
Quest Diagnostics	Client Services: 1-866-MYQUEST (697-8378)	Enzyme assay (Test Code: 38953)

Additional laboratories may also offer PK deficiency testing. For more information, visit [www.genetests.org](http://www.genetests.org) or [www.orpha.net](http://www.orpha.net).



[www.agios.com](http://www.agios.com)

- Zanella A, Fermo E, Bianchi P, Chiarelli LR, Valentini G. Pyruvate kinase deficiency: the genotype-phenotype association. *Blood Reviews* 2007; 21: 217-231.
- Hirono A, Kanno H, Miwa S, Beutler E. Chapter 182: Pyruvate Kinase Deficiency and Other Enzymopathies of the Erythrocyte. Valle D, Beaudet AL, Vogelstein B, et al, eds. *The Online Metabolic and Molecular Bases of Inherited Disease*. New York, NY: McGraw Hill; 2014. <http://ommbid.mhmedical.com/content.aspx?sectionid=62652268&bookid=971&Resultclick=2&q=PK+deficiency>. Accessed November 5, 2015.
- Grace RF, Zanella A, Neufeld EJ, Morton DH, Eber S, Yaish H, Glader B. Erythrocyte Pyruvate Kinase Deficiency: 2015 Status Report. *Am J Hematol*. 2015 Sep;90(9):825-30.