

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.^{1,2}

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



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
Thalassemias
Hereditary elliptocytosis

PK Deficiency

DIAGNOSTIC TESTS

1st

ENZYME ASSAY FOR PYRUVATE KINASE ACTIVITY

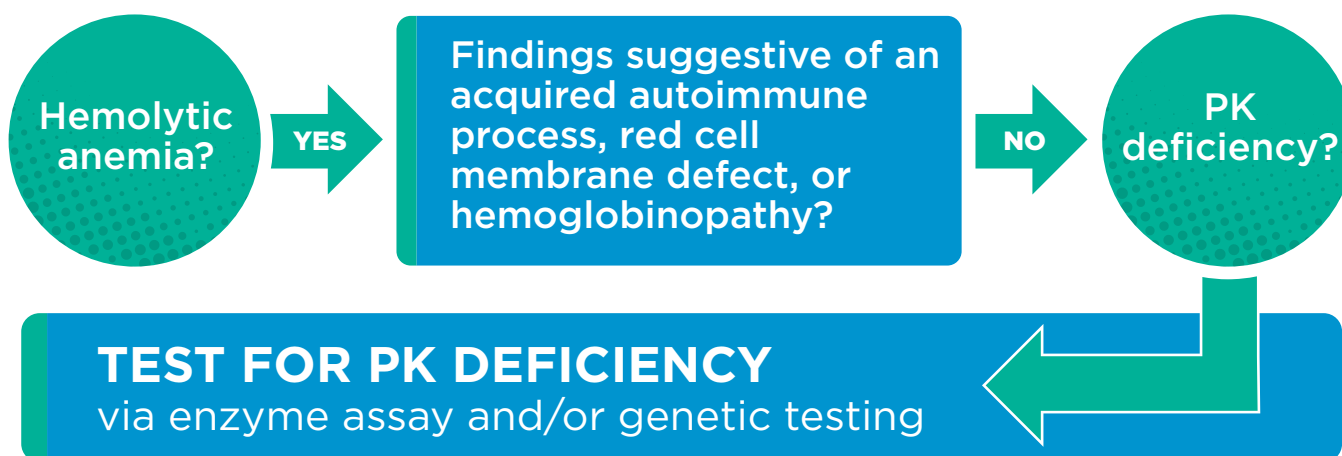
Enzyme assay is the gold standard for diagnostic testing of PK deficiency.³

2nd

MOLECULAR PK-LR ANALYSIS

Genetic testing may be conducted to confirm equivocal cases.³

TEST FOR PK DEFICIENCY



Diagnostic Testing for PK Deficiency

Laboratory	Contact Information	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 or www.orpha.net.

www.knowPKdeficiency.com



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- 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.