

ILLUMINATING

AN UNDERLYING CAUSE OF HEMOLYTIC ANEMIA

About PK Deficiency

OVERVIEW

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}

PK deficiency is the most common glycolytic defect causing hereditary nonspherocytic hemolytic anemia,^{1,3} yet may be underrecognized, particularly in adults and patients on the milder end of the spectrum of disease severity.²

PATIENT IDENTIFICATION

PK deficiency should be considered in patients with hemolysis and no findings suggestive of an acquired autoimmune process, red cell membrane defect, or hemoglobinopathy.³

Findings among PK-deficient patients may include:

- Anemia
- Splenomegaly
- Fatigue
- Gallstones
- Jaundice
- Iron overload

TREATMENT/MANAGEMENT

Current treatment is supportive and not disease-specific, and can include:

- Transfusion therapy
- Splenectomy
- Gall bladder removal (cholecystectomy)
- Iron chelation

Quick Facts

- Pyruvate kinase (PK) is a critical enzyme for maintaining red blood cell energy levels
- PK deficiency leads to decreased ATP and a shortened red cell lifespan
- Can be under- and misdiagnosed

ILLUMINATING PK DEFICIENCY

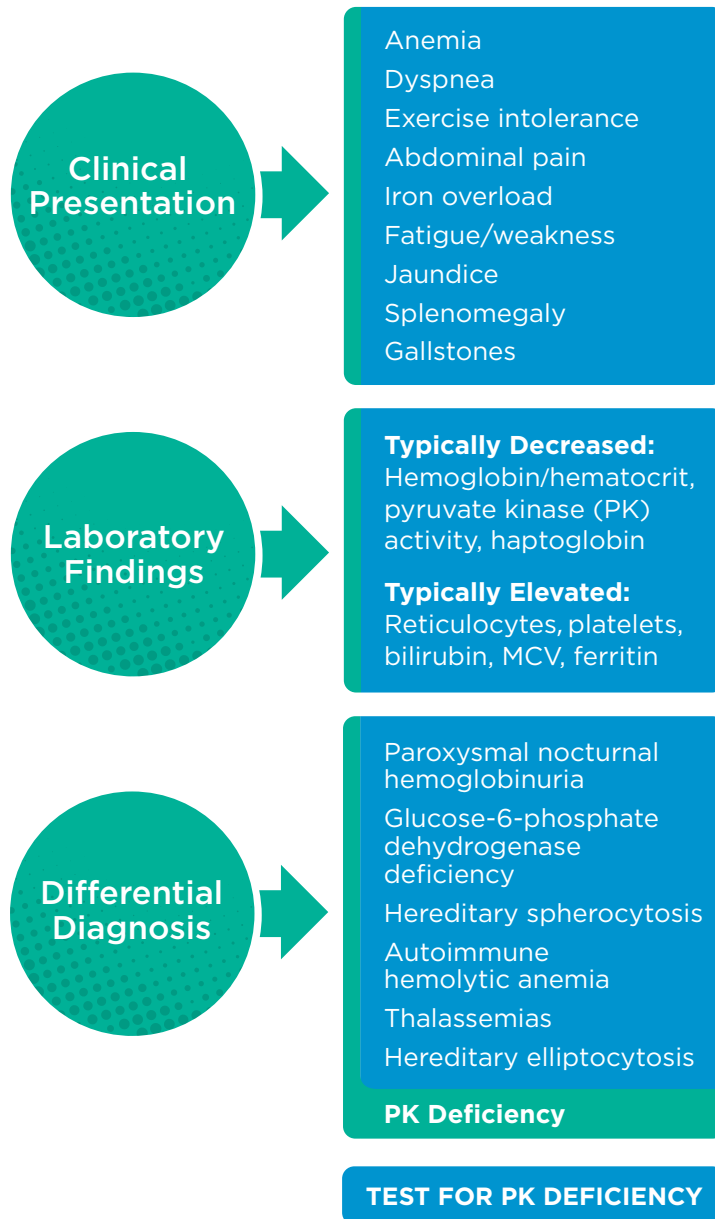
DIAGNOSIS

Enzyme assay (simple blood test) for pyruvate kinase activity should be conducted when PK deficiency is suspected.

PATIENT IMPACT

- Patient presentation ranges from mild to life-threatening^{1,2}
- Patients on the milder end of the spectrum may go years without a diagnosis²
- Fatigue, lethargy, and weakness may have a large impact on daily activity and quality of life

Proper patient identification and diagnosis are taking on new importance.



www.knowPKdeficiency.com



1. Zanella A, Fermo E, Bianchi P, Chiarelli LR, Valentini G. Pyruvate kinase deficiency: the genotype-phenotype association. Blood Reviews 2007; 21; 217-231.
2. Hirano 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.
3. 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.