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.¹²

PK deficiency is the most common glycolytic defect causing hereditary nonspherocytic hemolytic anemia,¹³ 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
- Fatigue
- Jaundice
- Splenomegaly
- Gallstones
- 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 diagnosis\(^2\)
• 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.

Clinical Presentation
- 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

Laboratory Findings

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

Differential Diagnosis

PK Deficiency

TEST FOR PK DEFICIENCY

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


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