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.\(^2\)

**Clinical Presentation**
- Anemia
- Dyspnea
- Exercise intolerance
- Abdominal pain
- Iron overload
- Fatigue/weakness
- Jaundice
- Splenomegaly
- Gallstones

**Laboratory Findings**
- Typically Decreased:
  - Hemoglobin/hematocrit
  - Pyruvate kinase (PK) activity
  - Haptoglobin
- Typically Elevated:
  - Reticulocytes
  - Platelets
  - Bilirubin
  - MCV
  - Ferritin

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

**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

<table>
<thead>
<tr>
<th>Laboratory</th>
<th>Contact Email and Phone</th>
<th>Type of Test</th>
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<tbody>
<tr>
<td>ARUP Laboratories, General Laboratory</td>
<td>Cynthia Gin, BS, MT (ASCP) <a href="mailto:ginca@aruplab.com">ginca@aruplab.com</a> 800-242-2787</td>
<td>Enzyme assay</td>
</tr>
<tr>
<td>Mayo Clinic, Metabolic Hematology Laboratory</td>
<td>Lea Koon, MS / Michelle Kluge, MS, CGC <a href="mailto:rstgchemepath@mayo.edu">rstgchemepath@mayo.edu</a> 800-533-1710</td>
<td>Enzyme assay</td>
</tr>
<tr>
<td>Cincinnati Children’s Hospital Medical Center, Molecular Genetics Laboratory (Cincinnati, OH)</td>
<td>Haley Keller / Chinmayee Nagaraj / Elizabeth Ulm / Emily Wakefield <a href="mailto:haley.keller@cchmc.org">haley.keller@cchmc.org</a> 513-636-4474</td>
<td>Enzyme assay</td>
</tr>
<tr>
<td>PreventionGenetics, Clinical DNA Testing and DNA Banking (Marshfield, WI)</td>
<td>Guoli Sun, MD, Ph, FACMG / Angela Gruber, PhD / Bruce Krawisz, MD <a href="mailto:clinicaltesting@preventiongenetics.com">clinicaltesting@preventiongenetics.com</a> 715-387-0484</td>
<td>Enzyme assay</td>
</tr>
<tr>
<td>Quest Diagnostics</td>
<td>Client Services: 1-866-MYQUEST (697-8378)</td>
<td>Enzyme assay, Genetic Testing (PKLR sequencing)</td>
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Additional laboratories may also offer PK deficiency testing. For more information, visit www.genetests.org or www.orpha.net.

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