Pyruvate Kinase Deficiency

Pyruvate kinase (PK) deficiency refers to a group of inherited disorders that result in a deficiency of the enzyme pyruvate kinase in the red blood cells. This enzyme is responsible for the conversion of phosphoenolpyruvate (PEP) to lactic acid in the liver and red blood cells. Deficiency of this enzyme leads to an accumulation of PEP in red blood cells, which can cause hemolytic anemia, weakness, fatigue, and other symptoms. There are different types of PK deficiency, each caused by a different mutation in the PKLR gene.

Test Details

**Indications for Use**
The Pyruvate Kinase Deficiency test is used to diagnose PK deficiency when clinical symptoms are present. The test is typically performed in patients with a family history of PK deficiency or when other tests such as a complete blood count (CBC) or glucose-6-phosphate dehydrogenase (G6PD) test are abnormal.

**Specific Considerations**
- PK deficiency is inherited in an autosomal recessive pattern, so family history is an important factor in considering the test.
- The test should be considered in patients with signs and symptoms suggestive of PK deficiency, such as hemolytic anemia, weakness, and fatigue.

**Test Performance Summary**
- The test is performed using a commercially available assay on an automated analyzer.
- The test has a turnaround time of 1-2 days.
- The test has a sensitivity and specificity of 95%.

**References**

**Change Log**
- **Date:** March 17, 2021
- **Change:** Updated Pyruvate Kinase Deficiency assay protocol.

Additional information.