HOTLINE: Effective November 18, 2019

New Test **3002059** **Pyruvate Kinase Deficiency (*PKLR*) Sequencing** **PKLR FGS**

Additional Technical Information

Out of Pocket Estimator

Patient History for Pyruvate Kinase Liver and RBC (*PKLR*) Sequencing Testing

Methodology: Polymerase Chain Reaction/Sequencing
Performed: Varies
Reported: 2-3 weeks

Specimen Required: Collect: Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Reference Interval: By report

Interpretive Data: Background information for Pyruvate Kinase Deficiency (*PKLR*) Sequencing:
Characteristics: Red cell pyruvate kinase (PK) deficiency, although relatively rare, is the most common glycolytic defect resulting in congenital non-spherocytic hemolytic anemia. Clinical features of PK deficiency are highly variable, ranging from well compensated anemia to severe disease with lifelong transfusion dependency. Other clinical manifestations may include jaundice, gallstones, iron overload and potential for other complications.
Prevalence: Varies by ethnicity; 1 in 20,000 Caucasians, higher prevalence in Pennsylvania Amish and Romani.
Inheritance: Autosomal recessive.
Cause: Pathogenic biallelic germline variants in *PKLR*.
Clinical Sensitivity: 98 percent.
Methodology: Bidirectional sequencing of all coding regions, intron/exon boundaries, 5' untranslated region and deep intronic variants c.1269+43T>C and c.1269+44C>T (also known as IVS9+43T>C and IVS9+44C>T, respectively) of the *PKLR gene*.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations or repeat element insertions. Deep intronic variants other than those targeted and large deletions/duplications will not be detected. Regulatory region variants outside of the 5' untranslated region will not be assessed.

See Compliance Statement C: www.aruplab.com/CS

CPT Code(s): 81405

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.