Test Definition: PKLRG

Pyruvate Kinase Liver and Red Blood Cell (PKLR), Full Gene Sequencing and Large Deletion Detection, Varies

Reporting Title: PKLR Full Gene and Deletion
Performing Location: Rochester

Ordering Guidance:

Preliminary screening tests, such as complete blood count with peripheral smear, direct Coombs test, and pyruvate kinase (PK) enzyme activity assays (preferably as a part of EEEV1 / Red Blood Cell [RBC] Enzyme Evaluation, Blood) should be run before ordering this evaluation.

Necessary Information:

1. PKLR Gene Sequencing Patient Information is required. Testing may proceed without the patient information however it aids in providing a more thorough interpretation. Ordering providers are strongly encouraged to complete the form and send it with the specimen.
2. Include physician name and phone number with specimen.

Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Yellow top (ACD solution B) or lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
Specimen Stability Information: Refrigerated 30 days

Specimen Type: DNA
Container/Tube: 2 mL screw top tube
Specimen Volume: 100 microliters
Collection Instructions:
1. The preferred volume is 100 microliters at a concentration of 250 ng/mcL
2. Include concentration and volume on tube
Specimen Stability Information: Frozen (preferred)/Ambient/Refrigerate

Specimen Minimum Volume:

Whole blood: 0.5 mL

Forms:

1. PKLR Gene Sequencing Patient Information is required
2. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)

3. If not ordering electronically, complete, print, and send a Benign Hematology Test Request Form (T755) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
37857	Result Summary	Alphanumeric		50397-9
48398	Result Details	Alphanumeric		82939-0
37858	Interpretation	Alphanumeric		69047-9
48397	Method	Alphanumeric		85069-3
48396	Disclaimer	Alphanumeric		62364-5
37860	Reviewed by	Alphanumeric		18771-6
91971	Additional Information	Alphanumeric		48767-8

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81405 81479

Reference Values:

An interpretive report will be provided.