

# 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

**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
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(PKLR), Full Gene Sequencing and Large  
Deletion Detection, Varies

Varies	Varies		
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Result Codes:

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