



Reporting Title: BPGM Full Gene Sequencing

Performing Location: Rochester

Ordering Guidance:

This test detects variants identifiable by Sanger sequencing in the BPGM gene only. For a reflexive evaluation including hemoglobin electrophoresis and variant analysis of genes associated with hereditary erythrocytosis, order REVE2 / Erythrocytosis Evaluation, Blood.

This test does not provide a serum erythropoietin (EPO) level. If EPO testing is desired, order EPO / Erythropoietin, Serum.

Necessary Information:

Erythrocytosis Patient Information (T694) is strongly recommended, but not required, to be filled out and sent with the specimen. This information aids in providing a more thorough interpretation of test results. Ordering providers are strongly encouraged to complete the form and send it with the specimen.

Specimen Requirements:

Submit only 1 of the following specimens:

Patient Preparation: Bone marrow transplants preclude accurate germline and genetic variant analysis. Inform the laboratory if this patient has undergone bone marrow transplantation. On rare occasions transfusion of blood products can preclude accurate genetic variant analysis, and results should be interpreted with caution if performed after recent transfusion (within 4 months).

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD), green top (sodium heparin)

Specimen Volume: 4 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in the original tube. Do not aliquot

Specimen Stability Information: Ambient 14 days (preferred)/Refrigerate 30 days

Specimen Type: Extracted DNA from whole blood

Container/Tube: 1.5 to 2 mL tube

Specimen Volume: Entire specimen

Collection Instructions:

1. Label specimen as extracted DNA and source of specimen
2. Provide volume and concentration of the DNA

Specimen Stability Information: Frozen (preferred)/Refrigerate/Ambient

Specimen Minimum Volume:

Blood: 1 mL; Extracted DNA: 50 mcL at 50 ng/mcL concentration



Forms:

1. 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:
 - Informed Consent for Genetic Testing (T576)
 - Informed Consent for Genetic Testing-Spanish (T826)
2. Erythrocytosis Patient Information (T694)
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®
37111	BPGM Gene Sequencing Result	Alphanumeric		No LOINC Needed
37112	BPGM Interpretation	Alphanumeric		69047-9

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81479

Reference Values:

An interpretive report will be provided.