



**MAYO**  
Mayo Medical Laboratories  
1-800-533-1710

## **NEW TEST ANNOUNCEMENT**

**NOTIFICATION DATE:** March 26, 2013

**EFFECTIVE DATE:** March 29, 2013

### **PPOX Gene, Known Mutation**

Test ID: PPOXK

**USEFUL FOR:** Predictive testing for variegate porphyria when a PPOX mutation has been identified in an affected family member

**METHODOLOGY:** Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing

**REFERENCE VALUES:** An interpretive report will be provided.

**SPECIMEN REQUIREMENTS:** **Specimen must arrive within 96 hours of collection.**

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts

Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 full T-75 or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.

2. Do not expose specimen to heat or direct sunlight.

3. Do not stack wet specimens.

4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

**NOTE:**

- Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
- New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

**SPECIMEN STABILITY INFORMATION:**

Specimen Type	Temperature	Time
Varies	Varies	

**CAUTIONS:**

- The identification of a disease-causing mutation in an affected family member is necessary before predictive testing for other family members can be offered. If a familial mutation has not been previously identified, order PPOXS/61261 PPOX Gene, Full Gene Analysis.
- Analysis is performed for the familial mutation provided only. This assay does not rule-out the presence of other mutations within this gene or within other genes that may be associated with porphyria.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Any error in the diagnosis or in the pedigree provided to us, including false paternity, could lead to erroneous interpretation of results.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

**CPT CODE:**

PPOX Gene, Full Gene Analysis

81479-Unlisted molecular pathology procedure

Fibroblast Culture for Genetic Testing

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

**DAY(S) SET UP:** Tuesday, 10 am

**ANALYTIC TIME:** 10 days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator  
Telephone: 800-533-1710