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**Reporting Title: PPOX Gene, Known Mutation**

**Performing Location: Rochester**

**Specimen Requirements:**

Forms:

1. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527).
2. 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 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

Specimen Type	Temperature	Time
Varies	Varies	

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
33805	Reason for Referral	Alphanumeric		In Process
33806	Result	Alphanumeric		In Process
33807	Interpretation	Alphanumeric		In Process
33808	Method	Alphanumeric		In Process
33809	Amendment	Alphanumeric		In Process
33810	Specimen	Alphanumeric		In Process
33811	Source	Alphanumeric		In Process
33812	Reviewed By	Alphanumeric		In Process
33813	Release Date	Alphanumeric		In Process

**CPT Code Information:**

PPOX Gene, Known Mutation  
 81479-Unlisted molecular pathology procedure  
 Fibroblast Culture for Genetic Testing  
 88233-Tissue culture, skin or solid tissue biopsy (if appropriate)  
 88240-Cryopreservation (if appropriate)

**Reference Values:**

An interpretive report will be provided