Reporting Title: PPOX Gene, Full Gene Analysis
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®
33794	Reason for Referral	Alphanumeric		In Process
33795	Result	Alphanumeric		In Process
33796	Interpretation	Alphanumeric		In Process
33797	Method	Alphanumeric		In Process
33798	Amendment	Alphanumeric		In Process
33799	Specimen	Alphanumeric		In Process
33800	Source	Alphanumeric		In Process
33801	Reviewed By	Alphanumeric		In Process
33802	Release Date	Alphanumeric		In Process

CPT Code: 1 × 81479

Reference Values:

An interpretive report will be provided