MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: January 4, 2013
EFFECTIVE DATE: January 4, 2013

GRHPR Gene, Known Mutation
Test ID: GRHKM

USEFUL FOR:

- Carrier testing of individuals with a family history of primary hyperoxaluria type 2
- Diagnostic confirmation of primary hyperoxaluria type 2 when familial mutations in the *GRHPR* gene have been previously identified
- Prenatal testing when 2 familial mutations in the *GRHPR* gene have been previously identified in an affected family member

METHODOLOGY: Polymerase chain reaction (PCR)/DNA sequence analysis or gene dosage analysis by multiplex ligation-dependent probe amplification (MLPA)

REFERENCE VALUES: An interpretive report will be provided.

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

Submit only 1 of the following specimens:

Specimen Type: Whole 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

NOTE: Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on the maternal specimen.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

NOTE:

- Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) 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 disease-causing mutations 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 GRHMS/50037 *GRHPR* 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 primary hyperoxaluria type 2.
- 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.
- In addition to disease-related probes, this test utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

CPT CODE:

81479-Unlisted molecular pathology procedure

Maternal Cell Contamination, Molecular Analysis
81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pretransplant recipient and donor germline testing, posttransplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample, and donor testing. Twin zygosity testing, or maternal cell contamination of fetal cells) (if appropriate)

Amniotic Fluid Culture for Genetic Testing
88235-Tissue culture for amniotic fluid (if appropriate)
88240-Cryopreservation (if appropriate)

Fibroblast Culture for Genetic Testing
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

DAY(S) SET UP: Monday 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