MAYO CLINIC
Mayo Medical Laboratories

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: December 3, 2010
EFFECTIVE DATE: December 3, 2010

AGXT Gene, Known Mutation #89916

USEFUL FOR:
- Carrier testing of individuals with a family history of primary hyperoxaluria type 1
- Diagnostic confirmation of primary hyperoxaluria type 1 deficiency when familial mutations have been previously identified
- Prenatal testing when 2 familial mutations have been previously identified in an affected family member

METHODOLOGY:
- Polymerase Chain Reaction (PCR)
- DNA Sequence Analysis and Gene Dosage Analysis by Multiplex Ligation- Dependent Probe Amplification (MLPA).

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:
Blood
Container/Tube: Lavender-top (EDTA) tube or yellow-top (ACD) tube
Specimen Volume: 3 mL of whole blood

Amniotic Fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL of amniotic fluid

Chorionic Villi
Container/Tube: Transport media in 15-mL tube
Specimen Volume: 20 mg of chorionic villus

NOTE: Specimen must arrive within 96 hours of draw.

NOTE: Prenatal Specimens: All prenatal specimens must be accompanied by a maternal blood specimen. **Due to the complexity of prenatal testing, consultation with the laboratory is required** for all prenatal testing.

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 #89915 "*AGXT* Gene, Full Gene Analysis."
- Analysis is performed for the familial mutation(s) 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 PH1.

- We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Predictive testing of an asymptomatic child is not recommended.
- 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.

LIST FEE: $191.20
One of the following test(s) will be added at an additional charge:
$273.00 for #82555 "DNA Sequence, Follow-up Analysis"
$273.00 for #89917 "AGXT Gene Large Deletion/Duplication"

For amniotic fluid specimens, the following test will be added at an additional charge:
$554.10 for #80334 "Amniotic Fluid Culture for Genetic Testing"

For chorionic villus specimens, the following test will be added at an additional charge:
$246.10 for #80333 "Fibroblast Culture for Genetic Testing"

CPT CODE:
"*AGXT* Gene, Known Mutation"
83891-Isolation or extraction of highly purified nucleic acid
83912-Interpretation and report

"*AGXT* Gene, Large Deletion/Duplication"
83900-Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
83909-Separation and identification by high-resolution technique (if appropriate)
83914 x 13-Mutation identification by enzymatic ligation or primer extension, single segment, each segment (if appropriate)

"DNA Sequence, Follow-up Analysis"
83892-Enzymatic digestion (if appropriate)
83894-Separation by gel electrophoresis (if appropriate)
83898-Amplification, target, each nucleic acid sequence (if appropriate)
83909 x 2-Separation and identification by high-resolution technique (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)

ANALYTIC TIME: 5 days **DAY(S) SET-UP:** Fridays, 10 am

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