MAYO CLINIC
Mayo Medical Laboratories

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

AGXT Gene, Full Gene Analysis #89915

USEFUL FOR:
- Confirming a diagnosis of primary hyperoxaluria type 1
- Carrier testing for individuals with a family history of primary hyperoxaluria type 1 in the absence of known mutations in the family

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

NOTE: Specimen must arrive within 96 hours of draw.

CAUTIONS:
- A small percentage of individuals who are carriers or have a diagnosis of PH1 may have a mutation that is not identified by this method (eg, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of PH1 disease. For carrier testing, it is important to first document the presence of a PH1-gene mutation in an affected family member.
- In some cases, DNA alterations of undetermined significance may be identified.
- 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.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- 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.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

LIST FEE: $1290.85

The following test will be added at an additional charge:
$273.00 for #89917 "AGXT Gene Large Deletion/Duplication"

TOTAL FEE: $1563.85

CPT CODE:
"*AGXT* Gene, Full Gene Analysis"
83891-Isolation or extraction of highly purified nucleic acid
83898 x 13-Amplification, target, each nucleic acid sequence
83909 x 26-Separation and identification by high-resolution technique
83912-Interpretation and report

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

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