MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: July 24, 2012
EFFECTIVE DATE: July 31, 2012

Krabbe Disease, Known Mutation
Test ID: GALCK

USEFUL FOR:

- Carrier testing of individuals with a family history of Krabbe disease when a mutation(s) has been identified in an affected family member
- Diagnostic confirmation of Krabbe disease when familial mutations have been previously identified

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequencing Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: **Specimen must arrive within 96 hours of collection.**
Submit only 1 of the following specimens:

Preferred:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
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

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

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

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

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 STABILITY INFORMATION:

Specimen Type	Temperature	Time
varies	varies	

CAUTIONS:

- The identification of a disease-causing mutation in an affected family member is necessary before testing can be offered for other family members. If a familial mutation has not been previously identified, order GALCS/60696 Krabbe Disease, Full Gene Analysis.
- Analysis is performed for the familial mutations 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 metabolic disease.
- 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.
- 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:
$355.00
One of the following test(s) will be added at an additional charge:
$289.60 for FUSEQ/82555 DNA Sequence, Follow-up Analysis
$231.40 for FUPCR/82554 Follow Up PCR

For amniotic fluid specimens, the following test will be added at an additional charge:
$587.80 for AFC/80334 Amniotic Fluid Culture for Genetic Testing

For chorionic villus specimens, the following test will be added at an additional charge:
$261.10 for FBC/80333 Fibroblast Culture for Genetic Testing

CPT CODE: Krabbe Disease, Known Mutation
83891-Isolation or extraction of highly purified nucleic acid
83912-Interpretation and report
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

Follow Up PCR
83894-Separation by gel electrophoresis (if appropriate)
83898-Amplification, target, each nucleic acid sequence (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: Thursday 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