

## **NEW TEST ANNOUNCEMENT**

**NOTIFICATION DATE:** May 31, 2012 **EFFECTIVE DATE:** May 31, 2012

# **Gaucher Disease, Known Mutation**

Test ID: GBAKM

#### **USEFUL FOR:**

- Diagnostic confirmation of Gaucher disease when familial mutations have been previously identified
- Carrier screening of at-risk individuals when a mutation in the *GBA* gene has been identified in an affected family member

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequence 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:**

• Specimen Type: 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

• **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

• **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:** 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: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

**Specimen Volume:** 5 blood spots

Specimen Stability Information: Ambient (preferred)/Refrigerated

#### **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 GBAMS/60711 Gaucher Disease, 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 metabolic disease.
- 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:** \$635.00

- For amniotic fluid specimens, the following test will be added at an additional charge: \$571.80 for #80334 Amniotic Fluid Culture for Genetic Testing \$1,206.80= Total List Fee
- For chorionic villus specimens, the following test will be added at an additional charge: \$261.10 for #80333 Fibroblast Culture for Genetic Testing \$896.10= Total List Fee

#### **CPT CODE**:

• Gaucher Disease, Known Mutation

83891-Isolation or extraction of highly purified nucleic acid

83892-Enzymatic digestion

83894-Separation by gel electrophoresis

83898-Amplification, target, each nucleic acid sequence

83909 x 4-Separation and identification by high-resolution technique

83912-Interpretation and report

• 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:** Friday; 2 PM **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