



**Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion,  
PCR  
Test ID: GALCS**

**USEFUL FOR:**

- Second-tier test for confirming a diagnosis of Krabbe disease
- Carrier testing for individuals with a family history of Krabbe disease in the absence of known mutations in the family

**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

**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:** 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:**

- This analysis does not exclude a diagnosis of atypical Krabbe disease due to saposin A deficiency.
- A small percentage of individuals who are carriers or have a diagnosis of Krabbe disease may have a mutation that is not identifiable by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of Krabbe disease.
- In some cases, DNA alterations of undetermined significance may be identified.
- 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:**

\$1,600.00

For skin biopsy specimens, the following test will be added at an additional charge:

\$261.10 for FBC/80333 Fibroblast Culture for Genetic Testing

\$1861.10 = Total List Fee

**CPT CODE:**

Krabbe Disease, Full Gene Analysis

83891-Isolation or extraction of highly purified nucleic acid

83894-Separation by gel electrophoresis

83898 x 20-Amplification, target, each nucleic acid sequence

83909 x 38-Separation and identification by high-resolution technique

83912-Interpretation and report

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