Test Definition: GALCW

Galactocerebrosidase, Leukocytes

Overview

Useful For

Diagnosis of Krabbe disease

Follow-up testing for evaluation of an abnormal newborn screening result for Krabbe disease

This test is **not intended for** carrier detection.

Genetics Test Information

This test provides diagnostic testing for patients with clinical signs and symptoms suspicious for Krabbe disease.

Enzyme testing for galactocerebrosidase is included in the diagnostic workup for infants following a positive newborn screen result for Krabbe disease.

Testing Algorithm

If the patient has abnormal newborn screening result for Krabbe disease, immediate action should be taken. Refer to the appropriate American College of Medical Genetics and Genomics Newborn Screening ACT Sheet.(1,2)

The following information is available:
-Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase
-Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase and Psychosine

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)
- Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase
- Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase and Psychosine

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD

Ordering Guidance

This test will not detect carrier status. For differentiating alterations from disease-causing variants in affected patients and for carrier detection in family members, molecular sequencing of the *GALC* gene is necessary. Order GALC / Krabbe Disease, *GALC* Gene Sequencing with Deletion/Duplication, Varies.

Shipping Instructions

For optimal isolation of leukocytes, it is recommended the specimen arrives refrigerate within 6 days of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Specimen Required

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot**.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Biochemical Genetics Patient Information (T602)

3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

4 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD	Refrigerated (preferred)	6 days	
	Ambient	6 days	

Clinical & Interpretive

Clinical Information

Krabbe disease (globoid cell leukodystrophy) is an autosomal recessive disorder caused by a deficiency of the enzyme, galactocerebrosidase (GALC). GALC facilitates the lysosomal degradation of psychosine (galactosylsphingosine) and 3

other substrates (galactosylceramide, lactosylceramide, and lactosylsphingosine causing severe demyelination throughout the brain. Krabbe disease is caused by variants in the *GALC* gene, and it has an estimated frequency of 1 in 100,000 births. Although rare, a few infants with an infantile Krabbe disease-like phenotype due to deficiency of saposin A have been found. Saposin-A is a sphingolipid activator protein that assists galactocerebrosidase in its action on galactosylceramide.

Severely affected infants typically present between ages 3 to 6 months with increasing irritability and sensitivity to stimuli. Rapid neurodegeneration including white matter disease follows, with death usually occurring by 2 years. Some individuals have later onset forms of the disease that are characterized by ataxia, vision loss, weakness, and psychomotor regression presenting anywhere from age 6 months to the seventh decade of life. The clinical course of Krabbe disease can be variable, even within the same family.

Newborn screening for Krabbe disease has been implemented in some states. The early (presymptomatic) identification and subsequent testing of infants at risk for Krabbe disease may be helpful in reducing the morbidity and mortality associated with this disease. While treatment is mostly supportive, hematopoietic stem cell transplantation has shown some success if performed early, prior to onset of neurologic damage.

Reduced or absent galactocerebrosidase in leukocytes can indicate a diagnosis of Krabbe disease; however, a number of alterations in the *GALC* gene have been identified that result in reduced galactocerebrosidase activity in vitro but do not cause disease. The biomarker, psychosine (PSY / Psychosine, Blood Spot or PSYR / Psychosine, Whole Blood or PSYCF / Psychosine, Spinal Fluid), has been shown to be elevated in patients with active Krabbe disease. Molecular sequencing of the *GALC* gene (GALC / Krabbe Disease, *GALC* Gene Sequencing with Deletion/Duplication, Varies) is necessary for differentiating alterations from disease-causing variants in affected patients and for carrier detection in family members.

Reference Values

> or =0.300 nmol/hour/mg protein
An interpretative report will be provided.

Interpretation

When abnormal results are detected, a detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional biochemical testing, and in vitro, confirmatory studies (enzyme assay, molecular analysis), name and phone number of key contacts who may provide these studies, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

Pseudodeficiency of galactocerebrosidase causes reduced enzymatic activity but does not cause disease.

A Krabbe disease phenotype can also be caused in very rare cases by the absence of a physiologically active sphingolipid activator protein, saposin A.

Enzyme levels may be normal in individuals who have undergone hematopoietic stem cell transplant.

Clinical Reference

1. Newborn Screening ACT Sheet [Decreased galactocerebrosidase, elevated psychosine] Krabbe Disease (infantile form). American College of Medical Genetics and Genomics; 2021. Updated May 2022. Accessed July 22, 2025. Available

at www.acmg.net/PDFLibrary/Krabbe-Infantile.pdf
2. Newborn Screening ACT Sheet [Decreased galactocerebrosidase, mildly elevated psychosine] Krabbe Disease (late-onset form). American College of Medical Genetics and Genomics; 2021. Updated May 2022. Accessed July 22, 2025. Available www.acmg.net/PDFLibrary/Krabbe-Later-Onset.pdf
3. Elliott S, Buroker N, Cournoyer JJ, et al. Pilot study of newborn screening for six lysosomal storage diseases using Tandem Mass Spectrometry. Mol Genet Metab. 2016;118(4):304-309
4. Matern D, Gavrilov D, Oglesbee D, Raymond K, Rinaldo P, Tortorelli S. Newborn screening for lysosomal storage disorders. Semin Perinatol. 2015;39(3):206-216
5. Orsini JJ, Escolar ML, Wasserstein MP, et al. Krabbe disease. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2000. Updated October 11, 2018. Accessed July 22, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1238/
6. Liao HC, Spacil Z, Ghomashchi F, et al. Lymphocyte galactocerebrosidase activity by LC-MS/MS for post-newborn screening evaluation of Krabbe disease. Clin Chem. 2017;63(8):1363-1369
7. Kwon JM, Matern DM, Kurtzberg J, et al. Consensus guidelines for newborn screening, diagnosis and treatment of infantile Krabbe disease. Orphanet J Rare Dis. 2018;13:30 doi:10.1186/s13023-018-0766-x

Performance

Method Description
The specimens are incubated with a mix of substrate and internal standard for galactocerebrosidase and alpha galactosidase (GLA). The reaction is then stopped using acetonitrile, centrifuged, and a portion of the supernatant is prepared for analysis by liquid chromatography-tandem mass spectrometry. GLA is included to verify sample integrity.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Preanalytical processing: Monday through Saturday
Testing performed: Monday, Wednesday

Report Available
3 to 7 days

Specimen Retention Time
White blood cell homogenate: 1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82657

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
GALCW	Galactocerebrosidase, WBC	24084-6

Result ID	Test Result Name	Result LOINC® Value
606270	Galactocerebrosidase, WBC	24084-6
606271	Interpretation	59462-2
606272	Reviewed By	18771-6