Test Definition: GALK

Galactokinase, Blood

Overview

Useful For

Diagnosis of galactokinase deficiency

Evaluation of children with unexplained bilateral congenital or juvenile onset cataracts

Genetics Test Information

Enzymatic testing for the diagnosis of galactokinase deficiency.

Testing Algorithm

For more information see Galactosemia Testing Algorithm.

Special Instructions

- Informed Consent for Genetic Testing
- Galactosemia Testing Algorithm
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)
- Galactosemia-Related Test List

Method Name

Enzyme Reaction followed by Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood EDTA

Ordering Guidance

This test is for diagnosis of galactokinase (GALK) deficiency and does **not** detect either galactose-1-phosphate uridyltransferase (GALT) deficiency (the most common cause of galactosemia), uridine diphosphate-galactose 4' epimerase (GALE) deficiency, or galactose mutarotase (GALM) deficiency. In most cases, GALT deficiency should be ruled out prior to evaluating for GALK deficiency.

-The preferred test to evaluate for possible diagnosis of galactosemia, routine carrier screening, and follow-up of abnormal newborn screening results is GCT / Galactosemia Reflex, Blood.

-To evaluate GALT deficiency only, order GALT / Galactose-1-Phosphate Uridyltransferase, Blood

-To evaluate for GALE deficiency only, order GALE / UDP-Galactose 4' Epimerase (GALE), Blood

-To evaluate for GALM deficiency, order GALP / Galactose, Plasma and molecular analysis of the *GALM* gene.

This assay is **not appropriate** for monitoring dietary compliance. If dietary monitoring is needed, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

Shipping Instructions

Pre-analytical processing is performed Monday through Friday and Sunday. This test may be canceled if specimens are outside of stability when processing occurs. Collect and package specimens for arrival on days when processing is performed.

Necessary Information

Biochemical Genetics Patient Information (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Required

Multiple whole blood tests for galactosemia can be performed on 1 specimen. Prioritize order of testing when submitting specimens. For a list of tests that can be ordered together see Galactosemia-Related Test List.

Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Green top (sodium or lithium heparin) or yellow top (ACD)
Specimen Volume: 4 mL

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

Specimen Minimum Volume

2 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated (preferred)	10 days	
	Ambient	72 hours	

Clinical & Interpretive

Clinical Information

Galactokinase (GALK) deficiency is a very rare autosomal recessive disorder in the first step of galactose metabolism. Prevalence is unknown but is estimated to be approximately 1 in 50,000-1 in 100,000 live births, with a higher frequency in the Romani population. Individuals with GALK deficiency have a milder clinical presentation than that seen in patients with classic galactosemia, galactose-1-phosphate uridyltransferase deficiency. The major clinical manifestation is bilateral juvenile cataracts.

GALK deficiency is treated with a galactose-restricted diet. Early treatment may prevent or reverse the formation of cataracts.

In GALK deficiency, erythrocyte galactose-1-phosphate levels are generally normal, and plasma or urine galactose levels are generally elevated. The diagnosis is established by demonstrating deficient GALK enzyme activity in erythrocytes. Testing for GALK deficiency should be performed when there is a suspicion of galactosemia, either based upon the patient's clinical presentation or laboratory studies and GALT deficiency has been excluded. Specimens sent for GALT analysis may be used for GALK testing if the original specimen was received in the laboratory within the stability parameters listed in Specimen Stability Information.

GALK deficiency is caused by variants in the *GALK1* gene. Molecular analysis of the *GALK1* gene is available; order CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies and specify Gene List ID: IEMCP-C2DU1U.

For more information see Galactosemia Testing Algorithm.

Reference Values

> or =0.7 nmol/h/mg of hemoglobin

Interpretation

An interpretive report will be provided.

Deficient galactokinase enzyme activity in erythrocytes is diagnostic for galactokinase deficiency.

Cautions

The results of testing performed in erythrocytes, including analysis of enzymes, biochemical phenotyping, or galactose-1-phosphate, are invalid following a transfusion.

Clinical Reference

1. Pasquali M, Yu C, Coffee B. Laboratory diagnosis of galactosemia: a technical standard and guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2018;20(1):3-11. doi:10.1038/gim.2017.172
2. Hennermann JB, Schadewaldt P, Vetter B, Shin YS, Monch E, Klein J. Features and outcome of galactokinase deficiency in children diagnosed by newborn screening. J Inherit Metab Dis. 2011;34(2):399-407. doi:10.1007/s10545-010-9270-8
3. Ramani PK, Arya K. Galactokinase deficiency. In: StatPearls [Internet]. StatPearls Publishing; 2021. Updated July 31, 2023. Accessed September 12, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK560683/
4. Walter JH, Fridovich-Keil JL. Galactosemia. In: Valle D, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019 Accessed September 12, 2024. Available

at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=%20225081023

Performance

Method Description
A buffered enzyme incubation with substrate and cofactors is performed on lysed red blood cells. A post-incubation extraction is performed and subjected to liquid chromatography tandem mass spectrometry. The ratio of the extracted product to its internal standard is used to calculate the total enzymatic product. This is then normalized using the calculated hemoglobin concentration to determine the patient's enzyme level in nmol/h/mg of hemoglobin.(Unpublished Mayo method).

PDF Report
No

Day(s) Performed
Preanalytical processing: Monday through Friday, Sunday
Assay performed: Monday

Report Available
5 to 11 days

Specimen Retention Time
2 months

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
82759

LOINC® Information

MAYO CLINIC LABORATORIES

Test Definition: GALK

Galactokinase, Blood

Test ID	Test Order Name	Order LOINC® Value
GALK	Galactokinase, B	81143-0

Result ID	Test Result Name	Result LOINC® Value
38005	Galactokinase, B	81143-0
38007	Interpretation (GALK)	59462-2
38006	Reviewed By	18771-6