Test Definition: GALK

Galactokinase, B

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 (GALK) deficiency.

Testing Algorithm

See Galactosemia Testing Algorithm in Special Instructions for additional information.

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

Advisory Information

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, or uridine diphosphate-galactose 4' epimerase (GALE) 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 followup 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

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

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. See Galactosemia-Related Test List in Special Instructions for a list of tests that can be ordered together.

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 in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Biochemical Genetics Patient Information(T602) is recommended, see Special Instructions.

3. If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism 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 and Interpretive

Clinical Information

Galactokinase (GALK) deficiency, is a very rare autosomal recessive disorder in the first step of galactose metabolism. Prevalence is unknown but 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 (GALT) 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. Gene analysis is available from some commercial laboratories. Call 800-533-1710 for recommendations or contact information for laboratories that offer this testing.

See Galactosemia Testing Algorithm in Special Instructions.

Reference Values

> or =0.7 nmol/h/mg of hemoglobin

Interpretation

An interpretive report will be provided.

Deficient galactokinase (GALK) enzyme activity in erythrocytes is diagnostic for galactokinase deficiency.

See Galactosemia Testing Algorithm in Special Instructions for additional information.

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. Li Y, Ptolemy AS, Harmonay L, et al: Ultra fast and sensitive liquid chromatography tandem mass spectrometry based assay for galactose-1-phosphate uridylyltransferase and galactokinase deficiencies. Mol Gen Metab 2011;102(1):33-40

2. Ko DH, Jun SH, Park HD, et al: Multiplex enzyme assay for galactosemia using ultraperformance liquid chromatography-tandem mass spectrometry. Clin Chem 2010;56:764-771

3. Hennermann JB, Schadewaldt P, Vetter B, et al: Features and outcome of galactokinase deficiency in children diagnosed by newborn screening. J Inherit Metab Dis 2011;34:399-407

4. Walter JH, Fridovich-Keil JL: Galactosemia. In *The Online Metabolic and Molecular Bases of Inherited Disease.* Edited by D Valle, AL Beaudet, B Vogelstein, et al. McGraw-Hill. Accessed June 18, 2019. Available at http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62672411

Performance

Method Description

A buffered enzyme incubation with substrate and cofactors is performed on lysed red blood cells. A postincubation 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) and Time(s) Test Performed

Mondays; 9 a.m.

Analytic Time

8 days

Maximum Laboratory Time

15 days

Specimen Retention Time

2 months

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

82759

LOINC® Information

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