Test Definition: HK1

Hexokinase Enzyme Activity, Blood

Overview

Useful For
Evaluating individuals with Coombs-negative chronic hemolysis

Method Name
Kinetic Spectrophotometry (KS)

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD-B

Specimen Required
Container/Tube:
Preferred: Yellow top (ACD solution B)
Acceptable: Yellow top (ACD solution A), Lavender top (EDTA)
Specimen Volume: 6 mL
Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot**.

Forms
If not ordering electronically, complete, print, and send a Benign Hematology Test Request (T755) with the specimen.

Specimen Minimum Volume
1 mL

Reject Due To

Gross hemolysis	Reject
Fully clotted	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated	20 days	

Clinical & Interpretive

Clinical Information

Hexokinase (HK) is the first enzymatic step in glycolysis, converting glucose to glucose 6-phosphate. Hexokinase deficiency (OMIM 235700) is a rare cause of chronic nonspherocytic hemolytic anemia, and its inheritance is autosomal recessive. Clinically significant HK deficiency manifests in early onset anemia with variable severity ranging from mild to severe. Some patients show neurologic impairment of which the mechanism is unclear.

Reference Values

> or =12 months: 0.7-1.7 U/g Hb
Reference values have not been established for patients younger than 12 months.

Interpretation

Clinical correlation or genetic confirmation may be required to establish hexokinase (HK) deficiency as a cause of hemolytic anemia, as the assayed activity level in confirmed cases can vary from markedly decreased to borderline normal levels due to a compensated increase in enzyme by reticulocytes. Comparison of HK activity levels to other red blood cell enzyme activity can be very useful. Heterozygotes have moderately decreased to low normal HK levels and are expected to be clinically unaffected.

Increased HK activity may be seen when reticulocytes are increased and is not supportive of a diagnosis of HK deficiency.

Cautions

Recent transfusion may mask the patient's intrinsic enzyme activity and cause unreliable results.

Hexokinase (HK) activity level can vary from markedly decreased to borderline normal levels in affected individuals due to a compensated increase in enzyme by reticulocytes. Comparison of HK activity levels to other red blood cell enzyme activity can be very useful.

Clinical Reference

1. Koralkova P, Mojzikova R, van Oirschot B, et al. Molecular characterization of six new cases of red blood cell hexokinase deficiency yields four novel mutations in HK1. Blood Cells Mol Dis. 2016;59:71-76
2. Koralkova P, van Solinge WW, van Wijk R. Rare hereditary red blood cell enzymopathies associated with hemolytic anemia-pathophysiology, clinical aspects and laboratory diagnosis. Int J Lab Hematol. 2014;36(3):388-397

Performance

Method Description

Hexokinase catalyzes the reaction of adenosine triphosphate (ATP) and glucose to glucose 6-phosphate (G6P) and adenosine diphosphate (ADP). In this assay the formation of G6P is measured by linking its further oxidation to 6-phosphogluconate to the reduction of nicotinamide adenine dinucleotide phosphate (NADP[+]) through the G6P dehydrogenase reaction. The increase in absorbance, which occurs as NADP(+) is reduced to NADPH, is measured spectrophotometrically at 340 nm on an automated chemistry analyzer.(Beutler E: Red Cell Metabolism: A Manual of Biochemical Methods. 3rd ed. Grune and Stratton; 1984:40-42; Rab MAE, van Wijk R. Enzymes of the red blood cell. In:

Rifai N, Chiu RWK, Young I, Burnham CAD, Wittwer CT, eds. Tietz Textbook of Laboratory Medicine. 7th ed. Elsevier; 2023:chap 78)

PDF Report
No

Day(s) Performed
Tuesday, Thursday

Report Available
1 to 6 days

Specimen Retention Time
7 days

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
HK1	Hexokinase, B	49216-5

Result ID	Test Result Name	Result LOINC® Value
HKCL	Hexokinase, B	49216-5