

Overview

Useful For

Evaluating individuals with chronic nonspherocytic hemolytic anemia

Evaluating individuals with early onset neurologic impairment

Genetic counseling for families with triosephosphate isomerase deficiency

Method Name

Only available as part of a profile. For more information see:

- HAEV1 / Hemolytic Anemia Evaluation, Blood
- EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Kinetic Spectrophotometry

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

Only available as part of a profile. For more information see:

- HAEV1 / Hemolytic Anemia Evaluation, Blood
- EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Specimen Minimum Volume

1 mL

Reject Due To

Gross hemolysis	Reject
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Specimen Stability Information

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

Clinical Information

Triosephosphate isomerase (TPI) converts dihydroxyacetone phosphate to glyceraldehyde 3-phosphate during glycolysis. Clinically significant TPI deficiency (OMIM #615512, autosomal recessive) is rare and classically manifests as a severe multisystem disorder with early hemolytic anemia and progressive neurologic impairment in infancy. Other clinical features include motor impairment, diaphragm paralysis, cardiomyopathy and susceptibility to infections. Some cases have isolated hemolytic anemia.

Reference Values

Only available as part of a profile. For more information see:

- HAEV1 / Hemolytic Anemia Evaluation, Blood
- EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

> or =12 months of age: 1033-1363 U/g Hb  
Reference values have not been established for patients who are younger than 12 months of age.

Interpretation

Clinically significant hemolytic anemias due to triosephosphate isomerase deficiency are associated with activity levels less than 30% of mean normal. Heterozygotes usually show approximately 50% of mean normal activity and are clinically unaffected.

Cautions

Recent transfusion may mask the enzyme activity of the patient and cause unreliable results.

Clinical Reference

1. Orosz F, Olah J, Ovadi J: Triosephosphate isomerase deficiency: facts and doubts. IUBMB Life. 2006 Dec;58(12):703-715. doi: 10.1080/15216540601115960

2. Fermo E, Bianchi P, Vercellati C, et al: Triose phosphate isomerase deficiency associated with two novel mutations in TPI gene. Eur J Haematol. 2010 Aug;85(2):170-173. doi: 10.1111/j.1600-0609.2010.01451.x

3. Tanaka KR, Zerez CR: Red cell enzymopathies of the glycolytic pathway. Semin Hematol. 1990;27:165

4. 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

Triosephosphate isomerase interconverts glyceraldehyde 3-phosphate and dihydroxyacetone phosphate (DHAP). The rate of DHAP formation is measured by further converting it to alpha-glycerophosphate by alpha-glycerophosphate

dehydrogenase which results in the oxidation of 1,4-dihydronicotinamide adenine dinucleotide (NADH) to NAD(+). The oxidation of NADH is measured spectrophotometrically by the decrease in absorbance at 340 nm on an automated chemistry analyzer. (Beutler E. Red Cell Metabolism. A Manual of Biochemical Methods. Grune and Stratton; 1984; van Solinge WW, van Wijk. Enzymes of the red blood cell. In: Rifai N, Horvath AR, Wittwer CT, eds. Tietz Textbook of Clinical Chemistry and Molecular Diagnostics. 6th ed. Elsevier; 2018:chap 30)

PDF Report

No

Day(s) Performed

Weekly, Varies

Report Available

5 days

Specimen Retention Time

7 days

Performing Laboratory Location

Rochester

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
TPIC	Triosephosphate Isomerase, B	44054-5

Result ID	Test Result Name	Result LOINC® Value
TPICL	Triosephosphate Isomerase, B	44054-5