Test Definition: TPI1

Triosephosphate Isomerase Enzyme Activity, Blood

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

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 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

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Triosephosphate Isomerase Enzyme Activity, Blood

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

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

> or =12 months: 1033-1363 U/g Hb

Reference values have not been established for patients younger than 12 months.

Interpretation

Clinically significant hemolytic anemias due to triosephosphate isomerase deficiency are associated with activity levels below 30% of mean normal. Heterozygous individuals 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;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;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(2):165-185

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

5. Williams A, Weisz-Hubshman M, Rossi V, et al. TPI deficiency: A case report and review of the literature. Mol Genet Metab. 2025;146(1-2):109227. doi:10.1016/j.ymgme.2025.10922

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. 3rd ed. Grune and Stratton; 1984; 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
TPI1	Triosephosphate Isomerase, B	44054-5

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