

Patient ID SA00143691	Patient Name VALIDATIONSOF, TESTING	Birth Date 1948-08-28	Gender F	Age 72
Order Number SA00143691	Client Order Number SA00143691	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 29 Mar 2021 18:00		

Triocephosphate Isomerase, B

Result Name	Value	Unit	Reference Value	Performing Site
  Triocephosphate Isomerase, B	500	U/g Hb	1033–1363	1 MCR

The Trioce-phosphate Isomerase (TPI) activity is decreased.

TPI activity levels are 50% of mean normal.

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

Clinically significant TPI deficiency is rare and classically manifests as a severe multisystem disorder with early hemolytic anemia and progressive neurologic impairment beginning in infancy. Other clinical features include motor impairment, diaphragm paralysis, cardiomyopathy and susceptibility to infections. Some cases with isolated chronic non-spherocytic hemolytic anemia without neurologic impairment have been reported.

If genotyping is desired, a sequencing panel is available. If indicated, please order NGHHA (Hereditary Hemolytic Anemia Seq) or NGENZ (RBC Enzyme Sequencing) and fill out the information sheet.

Received: 30 Mar 2021 14:40

Reported: 30 Mar 2021 14:41

Laboratory Notes

- 1** 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.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292