

Patient ID SA00143689	Patient Name VALIDATIONSOF, TESTING	Birth Date 1995-05-05	Gender M	Age 25
Order Number SA00143689	Client Order Number SA00143689	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 28 Mar 2021 08:00		

Phosphofructokinase, B

Result Name	Value	Unit	Reference Value	Performing Site
 Phosphofructokinase, B  Low	<1.2	U/g Hb	5.8–10.9	1 MCR

The Phosphofructokinase (PFK) activity level is decreased.

PFK activity level is 11% of mean normal range.

Clinically significant disorders due to PFK deficiency are associated with activity levels <50% of mean normal. Unaffected heterozygotes have been reported with levels of 63% of normal. Therefore genetic correlation may be helpful in ambiguous cases. Correlation with clinical features is necessary.

Clinically significant PFK deficiency, also called glycogen storage disease, type VII - Tarui disease (OMIM 232800, autosomal recessive) is a rare disorder with different clinical subtypes: classical, late-onset, infantile and hemolytic. Manifestations can variably include exercise intolerance, exertional myopathy, nausea, stiffness and myoglobinuria. Although not classically described, a second-wind effect is noticed by some patients (Sherman et al. PMID: 8037209). A subset of individuals have compensated hemolysis (normal hemoglobin values) or mild hemolytic anemia, episodic jaundice, hyperuricemia or gout-like symptoms.

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 14:35

Reported: 30 Mar 2021 14:37

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