

Patient Name SAMPLEREP,NEEVP A	Patient ID SA00059311	Age 47	Gender F	Order # SA00059311
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00059311	Account Information			Report Notes
Collected 06/24/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/25/2013 14:22				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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Neurologic Enzyme Evaluation
RECEIVED: 06/25/2013 12:22 **REPORTED:** 06/25/2013 13:16

Neurologic Enzyme Interpretation

MCR

Reviewed by KENNETH SWANSON

Triose-phosphate Isomerase (TPI) activity levels are 9% of mean normal. Clinically significant hemolytic anemias due to TPI deficiency are associated with activity levels <10% of mean normal. Heterozygotes usually show approximately 50% of mean normal activity and are hematologically normal. The heterozygote frequency has been estimated at 3-5% of the general population.

Clinically significant TPI deficiency is rare and manifests as a severe multisystem disorder with early hemolytic anemia and progressive neurologic impairment beginning before 14 months of age. Other clinical features include motor impairment, diaphragm paralysis, cardiomyopathy and susceptibility to infections.

Pyrimidine 5-Nucleotidase (P5NT) spectral absorption curve is abnormal indicating an increase in pyrimidine nucleotides. Lead inhibits P5NT activity; therefore heavy metal testing should be performed to exclude a reversible cause, otherwise in the correct clinical context, these results are supportive of P5NT deficiency.

Clinically significant P5NT deficiency is an autosomal recessive disorder that manifests as mild to moderate hemolytic anemia. Additional features are jaundice, splenomegaly and characteristic marked basophilic stippling on the peripheral blood smear. Coincident Hb E may lead to a more severe hemolytic anemia. Heavy Metal testing is available, if desired order Heavy Metals Screen with Demographics, Blood (test 15080 HMSBD), requires new sample, whole blood.

All other Red Blood Cell enzyme values are normal or elevated.

Glutathione, B	H	100.4	mg/dL RBC	46.9-90.1	MCR
Phosphofructokinase, RBC	H	10.0	U/g Hb	6.1 - 9.4	MCR
Phosphoglycerate Kinase, B	H	250	U/g Hb	165 - 239	MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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Triosephosphate Isomerase, B	L	115	U/g Hb	930 - 1406	MCR
Pyrimidine 5' Nucleotidase, B	AB	Abnormal			MCR
-- REFERENCE VALUE -- Expected result is normal					

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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