



Patient ID SA00059311	Patient Name SAMPLEREPORT, NEEVP A	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00059311	Client Order Number SA00059311	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Jun 2013 00:00		

Neurologic Enzyme Evaluation

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.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905



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Result Name	Value	Unit	Reference Value	Performing Site
 Glutathione, B	100.4	mg/dL RBC	46.9–90.1	MCR
 Phosphofructokinase, RBC	10.0	U/g Hb	6.1–9.4	MCR
 Phosphoglycerate Kinase, B	250	U/g Hb	165–239	MCR
 Triosephosphate Isomerase, B	115	U/g Hb	930–1406	MCR
 Pyrimidine 5' Nucleotidase, B	Abnormal			MCR

REFERENCE VALUE
Expected result is normal

Received: 25 Jun 2013 12:22

Reported: 25 Jun 2013 13:16

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905