

Patient ID SA00056621	Patient Name SAMPLEREPORT, TPPTL A	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00056621	Client Order Number SA00056621	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 Apr 2013 00:00		

TPP1 and PPT1, WBC

Reason for Referral

Not provided

MCR

Interpretation

MCR

In this sample the activity of Palmitoyl-Protein Thioesterase 1 is deficient and Tripeptidyl Peptidase 1 is normal. These results indicate that there is a defect in the CLN1 gene. Defects in this gene are associated with the clinical forms of Infantile, Late Infantile, Juvenile, and Adult neuronal ceroid lipofuscinosis. Molecular genetic testing of CLN1 is recommended for confirmation (see www.genetests.org for availability). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

Reviewed By

Marie Quade

MCR

Received: 17 Apr 2013 14:22


Reported: 17 Apr 2013 15:25

TPP1L

96 nmol/h/mg Prot

MCR

Reference Value
85-326

PPT1L
 1 nmol/h/mg Prot

MCR

Reference Value
20-93

Low

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