



Patient ID M0002260	Patient Name SAMPLEREPORT, TPPTF A	Birth Date 1971-09-28	Gender F	Age 40
Order Number M0002260	Client Order Number M0002260	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jun 2012 01:38		

TPP1 and PPT1, Fibroblasts

Reason For Referral

Not provided

TPP1, F



3 nmol/h/mg Prot

MCR

MCR

Reference Value
69–934

Interpretation

MCR

In this sample the activity of Tripeptidyl Peptidase 1 is deficient and Palmitoyl-Protein Thioesterase 1 is normal. These results indicate that there is a defect in the CLN2 gene. Defects in this gene are associated with clinical forms of Late Infantile and Juvenile neuronal ceroid lipofuscinosis. Molecular genetic testing of CLN2 is recommended for confirmation (see www.genetests.org for availability).

PPT1, F

36 nmol/h/mg Prot

MCR

Reference Value
30–194

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

MCR

Marie Quade

Received: 27 Jun 2012 01:38

Reported: 10 Apr 2013 11:32

Performing Site Legend

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