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|---|--|---|--------------------|------------------|
| 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.

TPP1L



96 nmol/h/mg Prot

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Reference Value
85-326

ADDITIONAL INFORMATION
Fluorometric Enzyme Assay

PPT1L

 1 nmol/h/mg Prot


MCR

Reference Value
20-93

Reviewed By

Marie Quade

MCR

Received: 17 Apr 2013 14:22

Reported: 17 Apr 2013 15:25

Performing Site Legend

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