



Patient ID C7028846-0005466	Patient Name TEST, NCLBS A	Birth Date 1972-01-31	Sex M	Age 51
Order Number X100426875	Client Order Number X100426875	Ordering Physician unkdr	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Aug 2023 23:15		

CLN1 and CLN2, BS

Reason for Referral

Follow up of known CLN1

Palmitoyl-protein thioesterase 1



2.05 nmol/mL/h

Low

Tripeptidyl peptidase 1

43.8 nmol/mL/h

Interpretation

POSITIVE In this dried blood spot sample, the activity of palmitoyl-protein thioesterase (PPT1) was reduced indicating that this individual may be affected with neuronal ceroid lipofuscinosis

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Reference Value
>10.0

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Reference Value
>27.0

1 MCR

1. The other enzyme analyzed by this method, tripeptidyl peptidase 1, was normal. PPT1 deficiency is associated with the clinical forms of infantile, late infantile, juvenile, and adult neuronal ceroid lipofuscinosis. For confirmation, we recommend molecular genetic analysis of the PPT1 gene (MCL test NCLGP). If not already ordered, this could be performed on the existing sample by calling MCL (1-800-533-1710) within one month. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Reviewed By

Marie Quade

MCR

Received: 14 Aug 2023 15:27

Reported: 14 Aug 2023 17:35

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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