

Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel,
Leukocytes

Patient ID C7028846-0005469	Patient Name TEST, NCLW A	Birth Date 1965-03-18	Sex F	Age 58
Order Number X100426877	Client Order Number X100426877	Ordering Physician unkdr	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Aug 2023 10:15		

CLN1 and CLN2, WBC

Reason for Referral

Rule out CLN2

Palmitoyl-protein thioesterase 1

30.6 nmol/h/mg Prot

Tripeptidyl peptidase 1

 10.6 nmol/h/mg Prot
Low

Interpretation

POSITIVE In this sample, the activity of tripeptidyl peptidase 1 (TPP1) in leukocytes was reduced indicating that this individual may be affected with neuronal ceroid lipofuscinosis 2. The other

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Reference Value
>10.0

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Reference Value
>20.0

1 MCR

enzyme analyzed by this method, palmitoyl-protein thioesterase, was normal. TPP1 deficiency is associated with clinical forms of late infantile and juvenile neuronal ceroid lipofuscinosis as well as autosomal recessive spinocerebellar ataxia-7 (SCAR7). For confirmation, we recommend molecular genetic analysis of the TPP1 gene (MCL test NCLGP). 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

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Received: 14 Aug 2023 15:28

Reported: 14 Aug 2023 18:31

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