MAYO CLINIC LABORATORIES

**NEW TEST**

Notification Date: September 18, 2023
Effective Date: October 17, 2023

# Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Leukocytes

**Test ID:** NCLW

## Useful for:

Supporting the biochemical diagnosis of two neuronal ceroid lipofuscinosis, CLN1 and CLN2 in whole blood specimens.

This is **not useful for** carrier detection.

## Ordering Guidance:

This blood test is an appropriate first step for individuals between 0 and 4 years of age who present with symptoms consistent with neuronal ceroid lipofuscinosis.

## Methods:

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

## Reference Values:

Palmitoyl-protein thioesterase 1: >10.0 nmol/h/mg protein

Tripeptidyl peptidase 1: >20.0 nmol/h/mg protein

An interpretative report will be provided.

## Specimen Requirements:

| | |
|---|---|
| **Preferred:** | Yellow top (ACD solution B) |
| **Acceptable:** | Yellow top (ACD solution A) or lavender top (EDTA) |
| **Specimen Volume:** | 6 mL |
| **Collection Instructions:** | Send whole blood specimen in original tube. **Do not aliquot.** |
| **Minimum Volume:** | 5 mL |

## Specimen Stability Information:

| Specimen Type | Temperature | Time |
|---|---|---|
| Whole Blood ACD | Refrigerated (preferred) | 6 days |
| | Ambient | 6 days |

## Cautions:

Deficiency of tripeptidyl peptidase 1 (TPP1) can also be indicative of autosomal recessive spinocerebellar ataxia-7.

Individuals with pseudodeficiency alleles can show reduced enzyme activity.

Carrier status (heterozygosity) for these conditions cannot be reliably detected.

Enzyme levels may be normal in individuals receiving enzyme replacement therapy or who have undergone hematopoietic stem cell transplant.

## CPT Code:

82657

## Day(s) Performed:

Preanalytical processing: Monday through Saturday
Testing performed: Tuesday

## Report Available:

8 to 15 days

## Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.