MAYO CLINIC LABORATORIES

NEW TEST

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

Neuronal Ceroid Lipofuscinosis, Two-Enzyme Panel, Blood Spot

Test ID: NCLBS

Useful for:
Supporting the biochemical diagnosis of two neuronal ceroid lipofuscinoses, CLN1 and CLN2

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

Genetics Information:
This test provides diagnostic testing for individuals with clinical signs and symptoms suspicious for neuronal ceroid lipofuscinosis 1 or 2 (CLN1 or CLN2). If an enzyme deficiency is detected by this screening test, additional biochemical or molecular testing is required to confirm a diagnosis.

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

Reference Values:
Palmitoyl-protein thioesterase 1: >10.0 nmol/mL/h

Tripeptidyl peptidase 1: >27.0 nmol/mL/h

An interpretative report will be provided.

Specimen Requirements:
Submit only 1 of the following specimen types:

Preferred:	
Specimen Type:	Blood spot
Supplies:	Card-Blood Spot Card Collection (Filter Paper) (T493)
Preferred:	Blood Spot collection Card
Acceptable:	Whatman Protein Saver 903 Paper, PerkinElmer 226 filter paper, Munktell filter paper, or blood collected in tubes containing ACD or EDTA and dried on filter paper.

Specimen Volume:	2 Blood spots
Collection Instructions:	1. An alternative blood collection option for a patient 1 year of age or older is a fingerstick. See How to Collect Dried Blood Spot Samples via fingerstick. 2. At least 2 spots should be complete (ie, unpunched). 3. Let blood dry on filter paper at room temperature in a horizontal position for a minimum of 3 hours. 4. Do not expose specimen to heat or direct sunlight. 5. Do not stack wet specimens. 6. Keep specimen dry.
Specimen Stability Information:	Refrigerated (preferred) 60 days/Ambient 7 days/Frozen 60 days
Additional Information:	1. For collection instructions, see Blood Spot Collection Instructions 2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777) 3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)
Minimum Volume:	1 Blood Spot

Acceptable:	
Specimen Type:	Whole blood
Preferred:	Lavender top (EDTA)
Acceptable:	Yellow top (ACD)
Specimen Volume:	2 mL
Collection Instructions:	Send whole blood specimen in original tube. **Do not aliquot.**
Specimen Stability Information:	Refrigerate (preferred) 7 days/Ambient: 48 hours
Minimum Volume:	0.5 mL

Specimen Stability Information:

Specimen Type	Temperature
Whole blood	Varies

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: Thursday **Report Available:** 8 to 15 days

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