

**Reporting Title:** CLN1 and CLN2, WBC  
**Performing Location:** Rochester

**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.

**Shipping Instructions:**  
For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 6 days of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

- Necessary Information:**
- 1. Patient's age is required.
  - 2. Reason for testing is required.

**Specimen Requirements:**  
**Container/Tube:**  
**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.**

- Forms:**
- 1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:  
[-Informed Consent for Genetic Testing \(T576\)](#)  
[-Informed Consent for Genetic Testing-Spanish \(T826\)](#)
  - 2. [Biochemical Genetics Patient Information \(T602\)](#)
  - 3. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request \(T798\)](#) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD	Refrigerated (preferred)	6 days	
	Ambient	6 days	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
NCLW	BG773	Reason for Referral: <ul style="list-style-type: none"><li>• Rule out CLN1</li><li>• Rule out CLN2</li><li>• Follow up of known CLN1</li><li>• Follow up of known CLN2</li></ul>	Answer List	Yes

		• Not Provided		
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Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
BG773	Reason for Referral	Alphanumeric		42349-1
618471	Palmitoyl-protein thioesterase 1	Numeric	nmol/h/mg Prot	74935-8
618472	Tripeptidyl peptidase 1	Numeric	nmol/h/mg Prot	76038-9
618473	Interpretation	Alphanumeric		59462-2
618470	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657

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.