

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.

Specimen Minimum Volume:

5 mL

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
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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">• Rule out CLN1• Rule out CLN2• Follow up of known CLN1• Follow up of known CLN2• Not Provided	Answer List	Yes

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.