



Reporting Title: CLN1 and CLN2, BS

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.

Necessary Information:

1. Patient's age is required.
2. Reason for testing is required

Specimen Requirements:

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

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 older than 1 year is a fingerstick. For detailed instructions, see How to Collect Dried Blood Spot Samples.
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)

Acceptable:

Specimen Type: Whole Blood

Container/Tube:

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

Specimen Minimum Volume:

Blood Spots: 1

Whole Blood: 0.5 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
Whole blood	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
NCLBS	BG757	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®
BG757	Reason for Referral	Alphanumeric		42349-1
618435	Palmitoyl-protein thioesterase 1	Numeric	nmol/mL/h	59246-9
618436	Tripeptidyl peptidase 1	Numeric	nmol/mL/h	72498-9
618437	Interpretation	Alphanumeric		59462-2
618434	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/mL/h

Tripeptidyl peptidase 1: >27.0 nmol/mL/h

An interpretative report will be provided.