

---

**Reporting Title:** Congenital Lactic Acidosis Panel**Performing Location:** Rochester**Ordering Guidance:**

The recommended first-tier tests to screen for an underlying biochemical etiology for congenital lactic acidosis (CLA) are a combination of the following:

Lactic acid in blood

LASF1 / Lactic acid, Spinal Fluid

ACRN / Acylcarnitines, Quantitative, Plasma

OAU /Organic Acids Screen, Random, Urine

AAQP / Amino Acids, Quantitative, Plasma

PDHC / Pyruvate Dehydrogenase Complex, Fibroblasts

Pyruvate carboxylase activity

Customization of this panel and single gene analysis for any gene present on this panel is available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

**Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

**Specimen Requirements:**

**Patient Preparation:** A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

**Specimen Type:** Whole blood

**Container/Tube:**

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. Invert several times to mix blood.

2. Send specimen in original tube. Do not aliquot.

**Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated 14 days

**Specimen Type:** Skin biopsy

**Supplies:** Fibroblast Biopsy Transport Media (T115)

**Container/Tube:** Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

**Specimen Volume:** 4-mm punch

**Specimen Stability Information:** Refrigerated (preferred)/Ambient

**Additional Information:** A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblast

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Blood spot

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

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card

Specimen Volume: 5 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. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.
2. For collection instructions, see Blood Spot Collection Instructions
3. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
4. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

### Specimen Minimum Volume:

See Specimen Required

### 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 in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing (Spanish) (T826)

2. Molecular Genetics: Biochemical Disorders Patient Information (T527) in Special Instructions

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

**Ask at Order Entry (AOE) Questions:**

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
608632	Test Description	Alphanumeric		62364-5
608633	Specimen	Alphanumeric		31208-2
608634	Source	Alphanumeric		31208-2
608635	Result Summary	Alphanumeric		50397-9
608636	Result	Alphanumeric		82939-0
608637	Interpretation	Alphanumeric		69047-9
608638	Resources	Alphanumeric		99622-3
608639	Additional Information	Alphanumeric		48767-8
608640	Method	Alphanumeric		85069-3
608641	Genes Analyzed	Alphanumeric		48018-6
608642	Disclaimer	Alphanumeric		62364-5
608643	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

**Supplemental Report:**  
Supplemental**CPT Code Information:**

81443  
81460  
81465  
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)  
88240-Cryopreservation (if appropriate)  
81479 (if appropriate for government payers)

**Reflex Tests:**

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes

**Result Codes for Reflex Tests:**

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

**Reference Values:**

An interpretive report will be provided.