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**Reporting Title:** Cystinuria Gene Panel**Performing Location:** Rochester**Ordering Guidance:**

The recommended first-tier test to screen for cystinuria is urinary measurement of cystine, lysine, ornithine, and arginine; order either CYSQN / Cystinuria Profile, Quantitative, 24 Hour, Urine or CYSR / Cystinuria Profile, Quantitative, Random, Urine.

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.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

**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. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD)

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send whole blood 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 Type:** Saliva  
**Patient Preparation:** Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.  
**Supplies:** Saliva Swab Collection Kit (T786)  
**Specimen Volume:** 1 Swab  
**Collection Instructions:** Collect and send specimen per kit instructions.  
**Specimen Stability Information:** Ambient 30 days  
**Additional Information:** Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing

- 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. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
608728	Test Description	Alphanumeric		62364-5
608729	Specimen	Alphanumeric		31208-2
608730	Source	Alphanumeric		31208-2
608731	Result Summary	Alphanumeric		50397-9
608732	Result	Alphanumeric		82939-0
608733	Interpretation	Alphanumeric		69047-9
608735	Additional Information	Alphanumeric		48767-8
608734	Resources	Alphanumeric		99622-3
608736	Method	Alphanumeric		85069-3
608737	Genes Analyzed	Alphanumeric		48018-6
608738	Disclaimer	Alphanumeric		62364-5
608739	Released By	Alphanumeric		18771-6

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

Supplemental Report:

Supplemental

CPT Code Information:

- 81479
- 88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
- 88240-Cryopreservation (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	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

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**Reference Values:**  
An interpretive report will be provided.