Test Definition: CYSQN

Cystinuria Profile, Quantitative, 24 Hour, Urine

Overview

Useful For

Diagnosis of cystinuria using 24-hour urine collections

Genetics Test Information

This test provides a biochemical diagnosis of cystinuria through the measurement of cystine, lysine, ornithine, and arginine.

Special Instructions

- Urine Preservatives-Collection and Transportation for 24-Hour Urine Specimens

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

1. **24-Hour volume (in milliliters) is required.**
2. **Patient's age is required.**
3. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.

Specimen Required

-Urine Tubes, 10 mL (T068)
-Diazolidinyl Urea (Germall) 5.0 mL (T822)
Specimen Volume: 5 mL
Collection Instructions:
1. Collect before intravenous pyelogram.
2. Add 5 g of diazolidinyl urea (Germall) as preservative **at start of collection**. If preservative is not available, refrigerate during collection.
3. Collect urine for 24 hours.
4. Mix well before taking 5-mL aliquot.
Additional Information: See Urine Preservatives-Collection and Transportation for 24-Hour Urine Specimens for multiple collections.

Forms

If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Urine Preservative Collection Options

Note: The addition of preservative must occur **prior to beginning the collection**. If preservative is not available, refrigerate during and after collection.

Ambient (no additive)	No
Refrigerate (no additive)	OK
Frozen (no additive)	OK
50% Acetic Acid	No
Boric Acid	No
Diazolidinyl Urea	Preferred
6M Hydrochloric Acid	No
6M Nitric Acid	No
Sodium Carbonate	No
Toluene	OK

Specimen Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	70 days	
	Refrigerated	14 days	

Clinical & Interpretive

Clinical Information

Cystinuria is an inborn error of metabolism resulting from poor absorption and reabsorption of the amino acid cystine in the intestines and kidneys. This leads to an accumulation of poorly soluble cystine in the urine and results in the production of kidney stones (urolithiasis). Symptoms may include acute episodes of abdominal or lower back pain and the presence of blood in the urine (hematuria). Recurrent episodes of kidney stones may result in frequent urinary tract infections, which may ultimately result in renal insufficiency. The combined incidence of cystinuria has been estimated to be 1 in 7000.

Cystinuria is an autosomal recessive disease, but some heterozygous carriers have an autosomal dominant, incomplete penetrance appearance with elevated, but typically nondisease-causing, urinary cystine excretion. Cystinuria is caused by variants in genes, *SLC3A1* on the short arm of chromosome 2 and *SLC7A9* on the long arm of chromosome 19.

Initially, the disease was classified into subtypes I, II, and III (type II and III are also referred as non-type I) based on the amount of urinary cystine excreted in heterozygous parental specimens. A new classification system has been proposed to distinguish the various forms of cystinuria: type A, due to variants in the *SLC3A1* gene; type B, due to variants in the *SLC7A9* gene; and type AB, due to 1 variant in each *SLC3A1* and *SLC7A9* gene.

Reference Values

Cystine
3-15 years: < or =53 mcmol/24 h
> or =16 years: < or =115 mcmol/24 h

Lysine
3-15 years: < or =140 mcmol/24 h
> or =16 years: < or =290 mcmol/24 h

Ornithine
3-15 years: < or =16 mcmol/24 h
> or =16 years: < or =70 mcmol/24 h

Arginine
3-15 years: < or =25 mcmol/24 h
> or =16 years: < or =64 mcmol/24 h

Conversion Formulas:
Result in mcmol/24 hours x 0.24 =result in mg/24 h
Result in mg/24 hours x 4.17 =result in mcmol/24 h

Interpretation

Individuals who are homozygous or compound heterozygous for cystinuria excrete large amounts of cystine in urine, but the amount varies markedly. Urinary excretion of other dibasic amino acids (arginine, lysine, and ornithine) is also typically elevated. Plasma concentrations are generally normal or slightly decreased.

Individuals who are homozygous and heterozygous for non-type I cystinuria can be distinguished by the pattern of urinary amino acids excretion: homozygous individuals secrete large amounts of cystine and all 3 dibasic amino acids, whereas heterozygous individuals secrete more lysine and cystine than arginine and ornithine.

Cautions

No significant cautionary statements

Clinical Reference

1. Servais A, Thomas K, Strologo LD, et al. Cystinuria: clinical practice recommendation. Kidney Int. 2021;99(1):48-58
2. Palacin M, Goodyer P, Nunes V, Gasparini P. Cystinuria. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw Hill; 2019. Accessed October 24, 2024. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225555540

Performance

Method Description
Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry. Patient samples are combined with isotopically labeled internal standard. Following protein precipitation, the supernatant is subjected to hydrophilic-interaction liquid chromatography for the separation of isomers with MS/MS detection of the underivatized amino acids.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
3 to 5 days

Specimen Retention Time
2 weeks

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82136

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CYSQN	Cystinuria Profile, QN, 24 hour	90785-7

Result ID	Test Result Name	Result LOINC® Value

TM38	Collection Duration	13362-9
VL36	Urine Volume	3167-4
3583	Cystine	13966-7
3589	Lysine	25464-9
3590	Ornithine	25491-2
3591	Arginine	25322-9
50447	Reviewed By	18771-6