Test Definition: CYSR

Cystinuria Profile, Quantitative, Random, Urine

Overview

Useful For

Biochemical diagnosis of cystinuria using random urine specimens

Genetics Test Information

This test provides biochemical diagnosis of cystinuria. It measures cystine, lysine, ornithine, and arginine.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

1. **Patient's age is required.**
2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)

Specimen Volume: 2 mL

Collection Instructions: Collect a random urine specimen.

Forms

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

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

Amino Acid		**Age groups**				
		<2 months	2-35 months	3-6 years	7-17 years	> or =18 years
Arginine	Arg	<250	<147	<81	<42	<65
Ornithine	Orn	<344	<238	<228	<291	<193
Cystine	Cys	<486	<285	<111	<84	<142
Lysine	Lys	<2217	<1321	<814	<463	<295

All results reported as nmol/mg creatinine.

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

MAYO CLINIC LABORATORIES

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Test ID	Test Order Name	Order LOINC® Value
CYSR	Cystinuria Profile, QN, Random	90784-0

Result ID	Test Result Name	Result LOINC® Value
23514	Cystine	30065-7
23515	Lysine	30048-3
23516	Ornithine	30049-1
23517	Arginine	30062-4
50416	Reviewed By	18771-6