Test Definition: MSUSC

Branched-Chain Amino Acids, Self-Collect, Blood Spot

Overview

Useful For

Monitoring patients with maple syrup urine disease using specimens collected at home

Special Instructions

- Blood Spot Collection Instructions-Fingerstick
- Blood Spot Collection Instructions-Fingerstick-Spanish

Highlights

This test is intended for dietary monitoring and follow-up of patients with maple syrup urine disease.

Method Name

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
Portions of this test are covered by patents held by Quest Diagnostics

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Necessary Information

Patient's street address, city, state, ZIP (postal) code, country, and home phone are required (post-office [PO] boxes are not acceptable delivery locations).

Specimen Required

Supplies: Blood Spot Collection-Self Collect (T858)
Container/Tube: Blood Spot Self Collection Card
Specimen Volume: 2 Blood spots
Collection Instructions:
1. Order test each time the patient is to collect a dried blood specimen at home and mail the specimen directly to Mayo Clinic Laboratories.
2. Order should be placed a minimum of 3 days prior to desired date of collection.
3. Enter patient's address information for each order created, including street address (**post-office [PO] boxes are not acceptable delivery locations**), city, state abbreviation, ZIP (postal) code, country, and home phone number.
4. For each order, the Blood Spot Collection-Self Collect kit will be mailed directly to the patient for self-collection (**delivery to a PO box will not occur**).

5. For more information on how to collect blood spots, see the following:
-How to Collect Dried Blood Spot Samples via fingerstick.
-Blood Spot Collection Instructions-Fingerstick
-Blood Spot Collection Instructions-Fingerstick-Spanish

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

Specimen Minimum Volume
1 Blood spot

Reject Due To

Blood spot specimen that shows serum rings or has multiple layers	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	59 days	FILTER PAPER
	Refrigerated	59 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER

Clinical & Interpretive

Clinical Information
Maple syrup urine disease (MSUD) is an inborn error of metabolism caused by the deficiency of the branched-chain-ketoacid dehydrogenase (BCKDH) complex. The BCKDH complex is involved in the metabolism of the branched-chain amino acids (BCAA): isoleucine (Ile), leucine (Leu), and valine (Val). Classic MSUD presents in the neonate with feeding intolerance, failure to thrive, vomiting, lethargy, and maple-syrup odor to urine and cerumen. If untreated, it progresses to irreversible intellectual disability, hyperactivity, failure to thrive, seizures, coma, cerebral edema, and possibly death.

Treatment of MSUD aims to normalize the concentration of BCAA by dietary restriction of these amino acids. BCAA are essential amino acids that require frequent adjustment of the dietary treatment. Dietary monitoring is accomplished by regular determination of BCAA and Allo-Ile concentrations.

Reference Values
Allo-isoleucine: <4 nmol/mL
Leucine: 52-269 nmol/mL

Isoleucine: 22-167 nmol/mL
Valine: 84-414 nmol/mL

Interpretation
Quantitative results of allo-isoleucine, leucine, isoleucine, and valine with reference values are reported without added interpretation.

Cautions
No significant cautionary statements

Clinical Reference
1. Morton DH, Strauss KA, Robinson DL, Puffenberger EG, Kelley RI. Diagnosis and treatment of maple syrup disease: a study of 36 patients. Pediatrics. 2002;109(6):999-1008. doi:10.1542/peds.109.6.999
2. Strauss KA, Puffenberger EG, Carson VJ. Maple syrup urine disease. In: Adam MP, Ardinger HH, Pagon RA, et al. eds. GeneReviews [Internet]. University of Washington, Seattle; 2006. Updated April 23, 2020. Accessed June 21, 2022. Available at www.ncbi.nlm.nih.gov/books/NBK1319/

Performance

Method Description
Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS). Patient samples are combined with isotopically labeled internal standard. Following extraction, the filtrate 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
2 to 4 days

Specimen Retention Time
1 year

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

0381U

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MSUSC	Branched-Chain Amino Acids, SC, BS	94571-7

Result ID	Test Result Name	Result LOINC® Value
618731	Allo-isoleucine	94572-5
618732	Leucine	47679-6
618733	Isoleucine	47671-3
618734	Valine	47799-2
618730	Reviewed By	18771-6
BG775	Patient Street Address (No PO Box)	56799-0
BG776	Patient City	68997-6
BG777	Patient State	46499-0
BG778	Patient Zip Code	45401-7
BG779	Patient Country	77983-5
BG780	Patient Home Phone	42077-8