

## 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:

- Order test each time the patient is to collect a dried blood specimen at home and mail the specimen directly to Mayo Clinic Laboratories.
- Order should be placed a minimum of 3 days prior to desired date of collection.
- 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.
- 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
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	59 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER
	Refrigerated	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: <2 nmol/mL  
Leucine: 35-215 nmol/mL

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Isoleucine: 13-130 nmol/mL

Valine: 51-325 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 Jun;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/](http://www.ncbi.nlm.nih.gov/books/NBK1319/)

### Performance

#### Method Description

This method quantifies valine (Val), allo-isoleucine (Allo-Ile), isoleucine (Ile), and leucine (Leu) using stable isotope-labeled internal standards (IS): d8-Val, d10-Allo-Ile, and d3-Leu. Branched-chain amino acids (BCAA) are extracted from a 3/16-inch dried blood spot (DBS) using a methanol:water (50:50) solution containing the IS. The filter plate containing the DBS and the IS are placed on an orbital shaker for 30 minutes at ambient temperature. The blood spot eluate is centrifuged into a 96-well round-bottom plate, dried under nitrogen, and reconstituted in aqueous mobile phase. BCAA are separated and detected by liquid chromatography-tandem mass spectrometry in positive selected reaction monitoring mode.(Oglesbee D, Kramer K, Lacey J, et al: Second-tier test for quantification of alloisoleucine and branched-chain amino acids in dried blood spots to improve newborn screening of maple syrup urine disease [MSUD]. *Clin Chem*. 2008 Mar;54(3):542-549. doi: 10.1373/clinchem.2007.098434; Stroek K, Boelen A, Bouva MJ, et al: Evaluation of 11 years of newborn screening for maple syrup urine disease in the Netherlands and a systematic review of the literature: Strategies for optimization. *JIMD Rep*. 2020 May 13;54(1):68-78. doi: 10.1002/jmd2.12124)

#### PDF Report

No

#### Day(s) Performed

Monday through Friday

#### Report Available

2 to 4 days

#### Specimen Retention Time

1 year

Performing Laboratory Location

Rochester

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