MAYO CLINIC LABORATORIES

Test Definition: AAMSD

Amino Acids, Maple Syrup Urine Disease Panel, Plasma

Overview

Useful For

Follow-up of patients with maple syrup urine disease

Monitoring of dietary compliance for patients with maple syrup urine disease

Highlights

This test is appropriate for follow-up and dietary monitoring 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

Plasma

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

Patient Preparation: Fasting (overnight preferred, 4 hours minimum). Infants should be drawn just before next feeding (2-3 hours without total parenteral nutrition: TPN if possible).
Collection Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Lavender top (EDTA), plasma gel tube, or green top (lithium heparin)
Submission Container/Tube: Plastic vial
Specimen Volume: 0.5 mL
Collection Instructions:
1. Collect specimen and place on wet ice. Note: Thrombin-activated tubes should not be used for collection.
2. Centrifuge immediately or within 4 hours of collection if specimen is kept at refrigerated temperature.
3. Being careful to ensure that no buffy coat is transferred, aliquot plasma into a plastic vial and freeze.

Forms

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

Specimen Minimum Volume

0.3 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen	14 days	

Clinical & Interpretive

Clinical Information

Maple syrup urine disease (MSUD) is an inborn error of metabolism caused by the deficiency of the branched-chain alpha-keto acid dehydrogenase (BCKDH) complex. The BCKDH complex is involved in the metabolism of the branched-chain amino acids (BCAA): isoleucine, leucine, and valine. MSUD can be divided into 5 phenotypes: classic, intermediate, intermittent, thiamine-responsive, and dihydrolipoyl dehydrogenase (E3)-deficient, depending on the clinical presentation and response to thiamin administration. Classic MSUD, the most common and most severe form, 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 diabilities, hyperactivity, failure to thrive, seizures, coma, cerebral edema, and possibly death.

Age of onset for individuals with variant forms of MSUD is variable and some have initial symptoms as early as 2 years of age. Symptoms include poor growth and feeding, irritability, and developmental delays. These patients can also experience severe metabolic intoxication and encephalopathy during periods of sufficient catabolic stress.

MSUD is a panethnic condition but is most prevalent in the Old Order Mennonite community in Lancaster, Pennsylvania with an incidence of 1:760 live births. The incidence of MSUD is approximately 1:185,000 live births in the general population.

Treatment of MSUD aims to normalize the concentration of BCAA by dietary restriction of these amino acids. Because BCAA are essential amino acids, the dietary treatment requires frequent adjustment, which is accomplished by regular determination of BCAA and allo-isoleucine concentrations. Orthotopic liver transplantation has been used with success and is an effective therapy for MSUD.

Reference Values

ISOLEUCINE
< or =23 months: 31-105 nmol/mL
2-17 years: 30-111 nmol/mL
> or =18 years: 36-107 nmol/mL

LEUCINE
< or =23 months: 48-175 nmol/mL
2-17 years: 51-196 nmol/mL
> or =18 years: 68-183 nmol/mL

VALINE
< or =23 months: 83-300 nmol/mL
2-17 years: 106-320 nmol/mL
> or =18 years: 136-309 nmol/mL

ALLO-ISOLEUCINE
< or =23 months: <2 nmol/mL
2-17 years: <3 nmol/mL
> or =18 years: <5 nmol/mL

Interpretation

The quantitative results of isoleucine, leucine, valine, and allo-isoleucine with age-dependent reference values are reported without added interpretation. When applicable, reports of abnormal results may contain an interpretation based on available clinical interpretation.

Cautions

Reference values are for fasting patients.

Clinical Reference

1. Chuang DT, Shih VE, Max Wynn RR. Maple syrup urine disease (Branched-chain ketoaciduria). 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 5, 2022. https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225084607
2. Frazier DM, Allgeier C, Horner C, et al: Nutrition management guideline for maple syrup urine disease: an evidence- and consensus-based approach. Mol Genet Metab. 2014 Jul;112(3)210-217. doi: 10.1016/j.ymgme.2014.05.006
3. Strauss KA, Puffenberger EG, Morton DH: Maple syrup urine disease. In: RA Pagon, MP Adam, HH Ardinger, et al, eds. GeneReviews[Internet]. University of Washington, Seattle; 2006. Updated April 23, 2020. Accessed October 5, 2022. Available at www.ncbi.nlm.nih.gov/books/NBK1319
4. Diaz VM, Camarena C, de la Vega A, et al: Liver transplantation for classical maple syrup urine disease: Long-term follow-up. J Pediatr Gastroenterol Nutr. 2014 Nov;59(5):636-639. doi: 10.1097/MPG.0000000000000469
5. Blackburn PR, Gass JM, Vairo FPE, et al:. Maple syrup urine disease: mechanisms and management. Appl Clin Genet. 2017 Sep 6;10:57-66. doi: 10.2147/TACG.S125962

Test Definition: AAMSD

Amino Acids, Maple Syrup Urine Disease Panel, Plasma

Performance

Method Description

Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS) by labeling amino acids present in plasma, urine, and spinal fluid with aTRAQ Reagent 121. Samples are dried and reconstituted with aTRAQ Reagent 113-labeled Standard Mix. Amino acids are separated and detected by LC-MS/MS. The concentrations of amino acids are established by comparison of their ion intensity (121-labeled amino acids) to that of their respective internal standards (113-labeled 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

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

82136

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AAMSD	Amino Acid, MSUD Panel, P	94566-7

Test Definition: AAMSD

Amino Acids, Maple Syrup Urine Disease Panel, Plasma

Result ID	Test Result Name	Result LOINC® Value
32446	Valine	94567-5
32447	Isoleucine	94568-3
32448	Leucine	94569-1
32449	Allo-isoleucine	94570-9
32450	Interpretation (AAMSD)	49247-0