Test Definition: TYRSC

Tyrosinemia Follow-Up Panel, Self-Collect, Blood Spot

Overview

Useful For

Monitoring of individuals with tyrosinemia type I (hepatorenal tyrosinemia) using a patient-collected specimen

Genetics Test Information

This test is intended for therapeutic monitoring of 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cycohexanedione (NTBC; nitisinone) and dietary monitoring of patients with tyrosinemia type 1.

Special Instructions

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

Highlights

Blood spot specimens for this test are self-collected by the patient to send directly to Mayo Clinic Laboratories via supplied collection kit. For more information, see Specimen Required.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Necessary Information

1. Patient's age is required.

2. **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

Container/Tube: Blood Spot Card

Specimen Volume: 2 Blood spots

Additional Information:

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 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 a Dried Blood Spot Sample 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
Insufficient specimen	Reject
Unapproved filter papers	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	7 days	FILTER PAPER
	Refrigerated	14 days	FILTER PAPER
	Frozen	90 days	FILTER PAPER

Clinical & Interpretive

Clinical Information
Tyrosinemia type 1 (hepatorenal tyrosinemia: HT-1) is an autosomal recessive condition caused by a deficiency of the enzyme fumarylacetoacetate hydrolase. HT-1 primarily affects the liver, kidneys, and peripheral nerves, causing severe liver disease, renal tubular dysfunction, and neurologic crises. If left untreated, most patients die of liver failure in the first years of life, and all are at risk of developing hepatocellular carcinoma (HCC). The incidence of HT-1 is approximately 1 in 100,000 live births.

Affected individuals can show a partial response to dietary restriction of phenylalanine and tyrosine, but dietary treatment in conjunction with the administration of 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3 cyclohexanedione (NTBC; nitisinone), an inhibitor of the proximal tyrosinemia pathway, is very effective when initiated in newborns. Outcome data are promising, and to date, newborn patients treated with NTBC have not developed acute liver disease, neurologic crises, or HCC.

According to treatment guidelines established in 2017, monitoring of blood NTBC concentration and succinylacetone (SUAC) levels, along with measuring the dietary intake of amino acids, including tyrosine and phenylalanine, are part of an individualized surveillance plan for patients with HT-1.(1) Monthly analysis of SUAC, NTBC concentration, and amino acids is suggested for the first year of life with the same compounds being monitored every 3 months to age 5 years and every 6 months thereafter.

The analytes encompassed in this assay satisfy the recommendations for diagnosing and monitoring HT-1. In particular, for NTBC, the current guidelines recommend 40 nmol/mL to 60 nmol/mL plasma concentration, which corresponds to a target range for NTBC in dried blood spots of 17 nmol/mL to 26 nmol/mL based on a blood to plasma conversion factor of 2.34.(2) Data from the validation of this assay suggests that NTBC dosing could be individualized while not to exceed DBS levels of 26 nmol/mL.(3)

Reference Values

Tyrosine:
<4 weeks: 40-280 nmol/mL
> or =4 weeks: 25-150 nmol/mL

Phenylalanine: 27-107 nmol/mL

Methionine: 11-45 nmol/mL

Succinylacetone: < or =1.0 nmol/mL

Nitisinone: < or =0.5 nmol/mL

Interpretation

Quantitative results with reference values are reported without added interpretation. When applicable, reports of abnormal results may contain an interpretation based on available clinical information.

Cautions

Bornaprine (Sormodrem) may, at least in theory, interfere with accurate measurement of 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3 cyclohexanedione (NTBC).

In rare cases of tyrosinemia type I, tyrosine or succinylacetone may not be elevated.

Clinical Reference

1. Chinsky JM, Singh R, Ficiciolglu C, et al. Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations. Genet Med. 2017;19(12). doi:10.1038/gim.2017.101
2. Laeremans H, Turner C, Andersson T, et al. Inter-laboratory analytical improvement of succinylacetone and nitisinone

quantification from dried blood spot samples. JIMD Rep. 2020;53(1):90-102
3. Mitchell GA, Grompe M, Lambert M, Tanguay RM. Hypertyrosinemia. 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 February 24, 2026. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225082825&bookid=2709#225082946
4. Blackburn PR, Hickey RD, Nace RA, et al. Silent tyrosinemia type I without elevated tyrosine or succinylacetone associated with liver cirrhosis and hepatocellular carcinoma. Hum Mutat. 2016;37(10):1097-1105. doi:10.1002/humu.23047
5. Ficicioglu C. Tyrosinemia type 1. In: Adam MP, Bick S, Mirzaa GM, et al, eds: GeneReviews [Internet]. University of Washington, Seattle; 2006. Updated November 20, 2025. Accessed December 30, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK1515/

Performance

Method Description
A 3-mm disk is punched out of the dried blood spot into a 96-well plate. The amino acids and nitisinone are extracted by the addition of acetonitrile and known concentrations of isotopically labeled amino acids as internal standards. The extract is moved to another 96-well plate, dried under a stream of nitrogen, and derivatized. Analytes are measured by liquid chromatography-tandem mass spectrometry. The concentrations of the analytes are established by computerized comparison of ion intensities of these analytes to that of the respective internal standards.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
3 to 5 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

0383U

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
TYRSC	Tyrosinemia Follow Up Panel, SC, BS	94573-3

Result ID	Test Result Name	Result LOINC® Value
610503	Tyrosine	35571-9
610504	Phenylalanine	29573-3
610505	Methionine	47700-0
610506	Succinylacetone	53231-7
610507	Nitisinone	85098-2
BG728	Reason for Referral	42349-1
610502	Reviewed By	18771-6
BG729	Patient Street Address (No PO Box)	56799-0
BG730	Patient City	68997-6
BG731	Patient State	46499-0
BG732	Patient Zip Code	45401-7
BG741	Patient Country	77983-5
BG733	Patient Home Phone	42077-8