
Reporting Title: Tyrosinemia Follow Up Panel, BS**Performing Location:** Rochester**Necessary Information:**

Patient's age and reason for testing are required.

Specimen Requirements:

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Blood Spot Collection Card

Acceptable: Whatman Protein Saver 903 Paper, PerkinElmer 226 filter paper, Munktell filter paper, or blood collected in tubes containing EDTA and dried on filter paper

Specimen Volume: 2 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect Dried Blood Spot Samples.
2. At least 2 spots should be complete (ie, unpunched).
3. Let blood dry on filter paper at room temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.

Specimen Stability Information: Ambient (preferred) 7 days/Refrigerated 14 days/Frozen 90 days

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

Acceptable:

Specimen Type: Whole blood

Container/Tube: Lavender top (EDTA)

Specimen Volume: 2 mL

Collection Instructions: Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Refrigerate 6 days

Specimen Minimum Volume:

Blood Spots: 1

Whole Blood: 0.5 mL

Forms:

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

Specimen Type	Temperature	Time	Special Container
Whole blood	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
TYRBS	BG722	Reason for Referral: <ul style="list-style-type: none">• Follow up of a known patient• Rule out Tyrosinemia Type I• Monitoring of treatment• Follow up of an abnormal newborn screen.	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
607553	Tyrosine	Numeric	nmol/mL	35571-9
607554	Phenylalanine	Numeric	nmol/mL	29573-3
607555	Methionine	Numeric	nmol/mL	47700-0
607556	Succinylacetone	Numeric	nmol/mL	53231-7
607557	Nitisinone	Numeric	nmol/mL	85098-2
BG722	Reason for Referral	Alphanumeric		42349-1
607552	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

84510
84030
82542
80299
82131
82542 only (if appropriate for government payers)

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