Lab Dept: Chemistry

Test Name: TYROSINEMIA FOLLOW UP PANEL, BLOOD SPOT

General Information

Lab Order Codes: TYRBS

Synonyms: Hereditary tyrosinemia, type I; Newborn screen follow up for elevated/normal tyrosine, elevated succinylacetone

CPT Codes:
84510 - Assay of Tyrosine
84030 - Phenylalanine, blood
82131 - Amino Acids; single, quantitative
82542 - Column chromatography, includes mass spectrometry, if performed, non-drug analyte not elsewhere specified, qualitative or quantitative
80299 - Quantitation of therapeutic drug, not elsewhere specified

Test Includes: Quantitative results for tyrosine, phenylalanine, methionine, succinylacetone, and nitisinone with reference values, reported without added interpretation. When applicable, reports of abnormal results may contain an interpretation based on available clinical information.

Logistics

Test indications: Useful for monitoring of individuals with tyrosinemia type I (HT-1).

Aids in the diagnosis of HT-1 when used in conjunction with testing for urine organic acids, liver function, alpha-fetoprotein, and molecular genetic analysis of the fumarylacetoacetate hydrolase (FAH) gene. This test assists in the diagnosis of tyrosinemia type 1 (HT-1) and monitoring of the effectiveness of 2-[2-nitro-4-trifluoromethylbenzoyl]-1,3-cycohexanedione (NTBC; nitisinone) and dietary therapy in patients with HT-1.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Clinic Laboratories (Mayo Test Code: TYRBS)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (performed by reference lab Monday through Friday)

Turnaround Time: 3-6 days

Special Instructions:	Contact the laboratory for blood spot filter paper collection card.

Specimen

Specimen Type:	Whole blood – blood spot
Container:	Card-Blood Spot Collection (Filter Paper) (Mayo supply T493 preferred) Alternative: Whatman Protein Saver 903 Paper, PerkinElmer 226 (formerly Ahlstrom 226) filter paper, Munktell filter paper, or blood collected in tubes containing EDTA and dried on filter paper.
Draw Volume:	2 blood spots filled (Minimum: 1 blood spot filled)
Processed Volume:	2 dried blood spots
Collection:	Warm the hand/heel being used for blood collection. Sterilize heel skin or finger with rubbing alcohol, dry, and puncture with sterile lancet not longer than 2 mm. Wipe away the first drop of blood with gauze. Allow large drops to form and apply directly to filter paper. Completely fill all circles with blood to allow saturation through the paper. Filled circles should appear the same on both sides of the paper. Send to lab to dry. • Do not squeeze tissue to obtain blood. • Do not use devices that contain EDTA or capillary tubes. • Do not apply specimen to both sides of filter paper.
Special Processing:	Allow blood to dry at room temperature in a horizontal position for 3 or more hours. Do not stack wet specimens or expose to heat. Forward promptly when dried, preferably within 24 hours of collection. Blood spot specimens stable at room temperature (preferred) for 7 days, refrigerated for 14 days, frozen for 90 days. See reference lab's catalog for alternative specimen type option.
Patient Preparation:	N/A
Sample Rejection:	Incompletely filled blood spots; layered blood spots; insufficient or multiple applications of blood spots; presence of serum rings; blood spots from specimens collected in EDTA; specimens collected on unapproved filter papers; unlabeled or mislabeled specimens.

Interpretive

Reference Range:	TYROSINE: < 4 weeks 40 – 280 nmol/mL ≥ 4 weeks 25 -150 nmol/mL PHENYLALANINE: 27 – 107 nmol/mL METHIONINE: 11 – 45 nmol/mL SUCCINYLACETONE: ≤ 1.0 nmol/mL NITISINONE: ≤ 0.5 nmol/mL
Critical Values:	N/A
Limitations:	Bornaprine (Sormodrem) may 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.
Methodology:	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
References:	Test Catalog - Mayo Clinic Laboratories (mayocliniclabs.com) (February 2024)
Updates:	2/14/2023: Initial entry (Note: Reference lab changed reference ranges on 2/13/23) 2/27/2024: Updated reference ranges and specimen stability.