

Indiana University School of Medicine
Department of Medical and Molecular Genetics
975 W. Walnut Street, IB 350, Indianapolis, IN 46202
Phone: 317-274-7597 Fax: 317-278-1616 CAP#1678930/CLIA#15D0647198

Affix label here

SHIP SPECIMENS TO: 975 W. Walnut Street, IB 350, Indianapolis, IN 46202

| PATIENT INFORMATION | | PHYSICIAN INFORMATION | |
|---------------------|------------------|-----------------------|---------------|
| Last Name | First Name | Ordering Physician | |
| Date of Birth | Sex | Institution Name | |
| Patient Email | | Physician Phone | Physician Fax |
| Patient Phone | Medical Record # | Physician Email | |

| REASON FOR REFERRAL | SPECIMEN INFORMATION | |
|---|---|---------------------------|
| <i>ICD-9 code(s) / Diagnosis / Other Relevant Clinic Notes:</i> | Date Collected | Time Collected |
| | Specimen Type <input type="checkbox"/> Plasma <input type="checkbox"/> Serum <input type="checkbox"/> Blood Spot <input type="checkbox"/> Urine <input type="checkbox"/> CSF | Collection Notes: |

TEST ORDER (Please circle/mark checkbox for your requested testing from the list below)

ACYLCARNITINE TESTS (only one box should be selected)

- 2000: Acylcarnitine Analysis, Basic [Plasma]**
- 4000: Acylcarnitine Analysis, Expanded [Plasma]**
- Targeted acylcarnitine subpanels (listed below)**
- If more than one is requested, TEST 4000 should be considered.*
- 4010: C3:DC / C4:OH Acylcarnitine [Plasma]
- 4040: C4:DC / C5:OH Acylcarnitine Isomers [Plasma]
- 4020: C4 Acylcarnitine Isomers [Plasma]
- 4030: C5 Acylcarnitine Isomers [Plasma]
- 4050: Long Chain Dicarboxylic Acylcarnitines [Plasma]
- 4060: Carnitine Metabolism Panel [Plasma]

ANALYTE TESTS: other

- 1100: Amino Acid Analysis [Plasma]**
- 1200: Amino Acid Analysis [Urine]**
- 1300: Amino Acid Analysis [CSF]
- 5000: Carnitine, Free and Total [Plasma]
- 5010: Methylmalonic Acid Analysis [Plasma or Serum]
- 1210: Orotic Acid Analysis [Urine]
- 3000: Organic Acid, Qualitative Analysis [Urine]
- 1400: Phenylalanine & Tyrosine Analysis [Blood Spot]

** See TEST DETAILS on next page for more information

| FOR LABORATORY USE ONLY | | | Specimen/order notes |
|-------------------------|----------------------|----------------------|----------------------|
| Date Received | Time Received | Received By | |
| <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |

Indiana University School of Medicine
Department of Medical and Molecular Genetics
975 W. Walnut Street, IB 350, Indianapolis, IN 46202
Phone: 317-274-7597 Fax: 317-278-1616 CAP#1678930/CLIA#15D0647198

SPECIMEN HANDLING: (Specimens received in lab Monday-Friday ONLY)

| | | |
|-------------------|---------------------|---|
| COLLECTION | PLASMA/SERUM | <ul style="list-style-type: none"> Separate plasma/serum from whole blood and freeze sample as soon as possible Samples stored as whole blood >4 hours are NOT ACCEPTABLE for amino acid analysis Gel-based separation tubes (PSTs or SSTs) are NOT ACCEPTABLE |
| | URINE | <ul style="list-style-type: none"> Collect random clean catch urine in a container free of preservatives or additives Freeze as soon as possible |
| | CSF | <ul style="list-style-type: none"> Plasma amino acid analysis should be collected and sent concurrently with all CSF amino acid analyses Collect CSF in a tube free of preservatives and store frozen as soon as possible CSF should appear clear and free of pink/yellow/red coloration Blood/plasma contaminated CSF samples will be analyzed but the result will likely be uninterpretable |
| | BLOOD SPOT | <ul style="list-style-type: none"> Use only Whatman 903 type blood spot cards Clean finger/heel prior to collection to remove lotions, soaps, powders or other foreign material Generate a single large blood drop prior to spotting. The goal is to fill a circle entirely with 1 drop. Allow cards to dry completely (at least 4 hours) prior to shipment. Visit our website (medicine.iu.edu/iubgl) for an instructional flyer on blood spot collection |

| | | |
|-----------------|--|--|
| SHIPPING | <p>SHIPPING ADDRESS:</p> <p>IU Biochemical Genetics Lab 975 W. Walnut Street, IB 350 Indianapolis, IN 46202</p> | <ul style="list-style-type: none"> A requisition form must be sent with each specimen Label all containers and forms with the patient's name, date of birth, MRN, and date of collection Ship frozen samples by overnight courier in an insulated container with 3 -5 lbs dry ice Serum methylmalonic acid and blood spot specimens may be shipped at ambient temperature if arriving within 1 week of collection Unused portions of specimens are stored in lab for a minimum of 2 months. |
|-----------------|--|--|

TEST DETAILS [Visit our website (medicine.iu.edu/iubgl) for more information including example reports]

AMINO ACID TESTING

- Analysis completed by LC-MS/MS using underivitized specimens.
- In addition to the traditionally covered amino acids, our panels contain multiple analytes not commonly included on amino acid assays therefore expanding the clinical utility to creatine metabolism disorders, sulfite intoxication diseases, and peroxisomal disorders
- Amino acids quantified in plasma: Alanine, Alloisoleucine, Alpha-aminoadipate, Alpha-amino-n-butyrate, Arginine, Argininosuccinate, Asparagine, Aspartate, Citrulline, Creatine, Creatinine, Glutamate, Glutamine, Guanidinoacetate, Glycine, Histidine, Homocitrulline, Homocystine, Hydroxyproline, Isoleucine, Leucine, Lysine, Methionine, Ornithine, Phenylalanine, Pimelic Acid, Proline, Sarcosine, Serine, Taurine, Threonine, Tryptophan, Tyrosine, Valine
- Amino acids quantified in urine: 3-Methylhistidine, Alanine, Alloisoleucine, Alpha-aminoadipate, Alpha-amino-n-butyrate, Anserine, Arginine, Argininosuccinate, Asparagine, Aspartate, Beta-alanine, Beta-Aminobutyrate, Citrulline, Creatine to Creatinine Ratio, Cystathionine, Cystine, Delta-aminolevulinic acid, Gamma-amino-n-butyrate, Glutamate, Glutamine, Guanidinoacetate, Glycine, Histidine, Homocitrulline, Homocystine, Hydroxyproline, Isoleucine, Leucine, Lysine, Methionine, Ornithine, Phenylalanine, Proline, Sarcosine, Serine, Sulfocysteine, Taurine, Threonine, Tryptophan, Tyrosine, Valine

ACYLCARNITINE TESTING

- TEST 2000 (Basic Acylcarnitine Analysis) is performed by flow injection tandem mass spectrometry of butylated specimens. This analysis is considered the traditional method for acylcarnitine testing and covers 33 common acylcarnitine species ranging from C2-C18 in chain length. This test is appropriate for diagnosis and management of numerous disorders of fatty acid oxidation and amino acid metabolism. LIMITATIONS: This method does not quantify free carnitine or resolve isobaric acylcarnitine species. Additional testing of free and total carnitine (TEST 5000) and urine organic acid analysis (TEST 3000) is sometimes needed to clarify findings.
- TEST 4000 (Expanded Acylcarnitine Analysis) is performed by liquid chromatography tandem mass spectrometry of underivitized specimens. This advanced method was developed by our laboratory as an improved alternative to current acylcarnitine testing (PMID 34954532). This test quantifies 61 analytes including all acylcarnitine species in TEST 2000 with the advantage of providing unambiguous separation and quantification of clinically important isomeric/isobaric acylcarnitines. This method also includes free carnitine, carnitine metabolic precursors, long chain dicarboxylic acylcarnitines useful in the diagnosis of peroxisomal disorders, and ratio calculations. LIMITATIONS: This test is completed biweekly and therefore has a longer turn-around-time compared to TEST 2000 which is completed multiple times each week.
- Targeted acylcarnitine panels (TESTs 4010-4060) are subpanels of TEST 4000. Specimens are subjected to TEST 4000 workflow but only the targeted analytes are analyzed and reported. These tests may be considered to follow-up on Newborn Screening results that do not clarify acylcarnitine isomer identity or when there is a high clinical concern for a specific metabolic disorder targeted by these panels. If multiple panels are needed, it is more cost effective to order TEST 4000.