Biochemical and Molecular Genetics Diagnostic Laboratory
University of Miami, Miller School of Medicine

JANUARY 2018
Manual of Service

1501 NW 10th Avenue
BRB, Room 535
Miami, Florida  33136
University of Miami, Biochemical and Molecular Genetics Diagnostic Laboratory

Table of Organization 2018

Mustafa Tekin, MD
Laboratory Director

Assistant Laboratory Director
Jingyu Huang, PhD

Manager and Compliance Officer
TBA

Laboratory Supervisor
TBA

Chief Financial Officer
Antonio Izquierdo

Revised
January 5, 2018
University of Miami, Miller School of Medicine

Mission Statement

- To provide excellence in medical education.
- To expand medical knowledge through research.
- To provide high-quality care to those who need it.
- To be a community partner.
Biochemical and Molecular Genetics Diagnostic Laboratory
University of Miami, Miller School of Medicine

General Information

Laboratory Director

Mustafa Tekin, MD  (305)-243-2381
Professor, Department of Human Genetics
MTekin@miami.edu

Team Members

Jingyu Huang, PhD  (305)-243-4856
Assistant Professor and Assistant Lab Director
JHuang@med.miami.edu

Belkis Alfonso, BS, MT (AMT)  (305)-243-5450
Medical Technologist
MRodriguez28@med.miami.edu

Mayra Rodriguez, MS, MT (ASCP),(AAB)  (305)-243-6671
Medical Technologist
MRodriguez28@med.miami.edu

Anel Frederick, BS
LIS Systems Analyst
AFrederick@med.miami.edu

Billing Officer
Deborah Paul  (305)-243-6583
Manager, Medical Billing & Collections
Geneticsbillingoffice@med.miami.edu

Lab Contact Numbers (305)-243-5450 phone  /  (305)-243-5451 fax
Client Support Services
Client Support Service is available for all of our clients including: physician offices, clinics, hospitals and all UM medical campus facilities. The address is noted below:

1501 NW 10th Avenue  
BRB, room 535  
Miami, FL 33136  
(305) 243-6671 or (305)-243-5450  
biochemgenlab@med.miami.edu

The Hours of operation are noted below in Eastern Standard Time:  
Monday to Friday 8:30 am to 5:00 pm

Introduction
The Biochemical and Molecular Genetics Diagnostic Laboratory is directed and closely supervised by members of the faculty of the University of Miami, Department of Human Genetics. The laboratory offers the highest quality laboratory assays (metabolic disorders and DNA sequencing) to area hospitals, physicians and other reference laboratories. In addition, geneticists (who are members of the UM faculty) are continuously involved in the development of new laboratory test procedures with unique clinical applications.

The staff of the Biochemical and Molecular Genetics Diagnostic Laboratory (BMDGL) is dedicated to the skilled handling and expert processing of laboratory specimens. Specific questions concerning specimen requirements, special handling procedures and assay results should be directed to BMDGL personnel at (305) 243-5450. Alternatively, our clients may contact the appropriate Client Support Services Office for issues associated with specimen transportation and/or clients may obtain necessary information by reading this document.

Billing
Clients should contact the Billing office for invoice inquiries. The BMDGL can only receive payment in US dollars. Clients may provide a draft from a financial institution with a US address.

Professional Consultation
Both the laboratory director and assistant director are available for consultation on problem cases and to discuss test results and their interpretation. The BMDGL only performs the clinical tests that are listed in this document.

Specimen Preparation
Specimen requirements for individual tests, which have critical handling demands, are noted under the test name of the request form. All containers must be properly labeled with the patient’s name, DOB, ordering physician name and the date and time of specimen collection. Please close and seal all containers properly to prevent unnecessary exposure to potentially infectious specimens. Specimens must be transported in an appropriate (biohazard) specimen bag. Laboratory request forms must accompany the specimen in an adjacent plastic bag to avoid contamination of request form. In the event a specimen received by BMDGL personnel is unsuitable and/or inappropriate for the requested laboratory analysis, we shall notify the client and or the requesting physician as soon as possible and return the specimen the following business day (please see specimens returned).

Note** Physician electronic orders and or written physician orders for laboratory testing, may be attached to the lab request form and will be accepted in lieu of signed lab request form.

Note** The PRIMARY SPECIMEN container must be labeled with 2 identifiers at the time of collection. (Examples of identifiers include but are not limited to patient name, DOB, hospital number, social security or medical record number, requisitions number, accession number, unique random number. A hospital room number is not an acceptable identifier).

Note ** The Biochemical and Molecular Genetics Diagnostic Laboratory (BMDGL) does not perform specimen collection. Specimen collection must be performed per client policies and procedures for patient identification, preparation and collection of specimen.

Specimen Transportation
The courier personnel are specially trained to ensure the proper handling and preservation of all types of specimens.

INTERNAL COURIER SERVICES
Internal courier transport services are available for clients/physicians located on the UM, Medical Campus. To arrange for specimen pick-up, please call Client Support Services phone number at (305) 243-5450.

EXTERNAL COURIER SERVICES
External courier transport services are available for clients/physicians that are NOT located on the UM, Medical Campus. To arrange for specimen pick-up, please call BMDGL phone number at (305)-243-5450.

Specimens emanating from outside of the South Florida tri-county area must be transported by the client via a courier or delivery company (i.e. FedEx) of their choice. BMGDL will not be responsible for the specimen until it is received by the laboratory.

All laboratory request forms are “clocked in” documenting the time the specimen is received by the BMGDL personnel. The “clocked in” time and date is the official time of specimen receipt. Test turn-around-time is based on the official time of specimen receipt.

Unacceptable Specimens
- Needles: We will not accept, transport or test any specimen that has or may contain an attached needle.
- Glass tubes: Glass transfer tubes should be avoided (if possible) due to the increased risk of broken glass and biological contamination.
- Improperly Labeled Specimens: We will not accept specimens submitted without proper identification; patient name, identification number, age, sex, date of birth, ordering physician, diagnosis and ICD-10 code, physician number, specimen type, date and time of collection, test request, and a completed laboratory request form which includes a patient consent form.

Patient Consent
The ordering physician assumes responsibility for obtaining appropriate patient (or guardian) consent for genetic testing. This includes family testing (i.e. testing for pre-symptomatic or asymptomatic individual for a known familiar mutation to determine disease pre-disposition and/or carrier status).

With reference to testing an affected individual, the laboratory will accept a signed laboratory request form or a written statement (i.e. email, letter or fax) from the physician. The ordering physician is responsible for genetic testing and has discussed the subject with the patient and/or legal guardian.

Note** Physician electronic orders and or written physician orders for laboratory testing, may be attached to the lab request form and will be accepted in lieu of signed lab request form

In the event the patient requires his/her specimen be destroyed after clinical testing is performed; it is the responsibility of the ordering physician to include that information on the lab request form. The BMGDL lab will destroy the specimen 60 days post results reporting.

Laboratory results will only be released to ordering physicians or health care providers (i.e. genetic counselors) for whom a duplicate report request is documented on the lab request form.

STAT
Stat work will be performed on a case by case basis. Physicians should contact the lab director to discuss the specifics of the testing that is required. Refer to the General Information page for contact numbers.

Specimen Returns
- Biochemical and Molecular Genetics Diagnostic Laboratory (BMGDL) will charge the client’s account a $15.00 handling fee to cover the cost of returning specimens, if a return is requested by the client.
- Requests must be made by the submitting laboratory in writing.
- FedEx account number may be provided in conjunction with request. Allow 2-3 business days for return of specimens.

NOTE** The Biochemical and Molecular Genetics Diagnostic Laboratory (BMGDL) will not be held responsible for returned specimens that may be damaged or lost during return transit.

Test Cancellations, Additions, STAT
Requests

- Tests may be cancelled by our Clients without charge while specimens are in transit to the laboratory. It will be the responsibility of the client to call our lab and send us a cancellation request in writing via fax or FedEx.
- Test cancellations cannot be made after an assay has been set-up or completed and will therefore be charged the standard test fee.
- Additional testing can be arranged if sufficient specimen volume permits.
- The Biochemical and Molecular Genetics Diagnostic Laboratory requires that requests for cancellation or additional testing be made by the originating / authorized laboratory.
- For all verbal requests, a written authorization must be submitted to BMGDL lab personnel within 48 hours (required for compliance with federal regulations).
- Requests for STAT pick-up and delivery of specimens may be subject to an additional charge.
- Requests must be made by the submitting laboratory in writing (i.e. all specimens must be accompanied by BMGDL request form).
- FedEx account number may be provided in conjunction with request (allow 2-3 business days for return of specimens), if a client account number has not been established.

Test Changes

The Biochemical and Molecular Genetics Diagnostic Laboratory continually strives to enhance services and improve the quality of laboratory test results. In so doing, turn-around-times, methodologies, reference ranges and prices are subject to change.

In the event of test delays or test changes (i.e. reference ranges) the client will be notified via a comment on the laboratory test result(s) (i.e. corrected reference ranges are...), newsletter, and individual contact with ordering physician. Notifications shall always be performed per Laboratory Director’s (or designee, i.e.: supervisor) instructions.

CPT Coding

The Biochemical and Molecular Genetics Diagnostic Laboratory has provided CPT Codes in an effort to offer some guidance. The CPT codes listed only reflect our interpretation. It is the client’s responsibility to determine the correct CPT to use for billing and verify the accuracy of the codes for the tests actually performed. The Biochemical and Molecular Genetics Diagnostic Laboratory assumes no responsibility for billing errors due to the clients reliance on the CPT codes listed in this document. For further reference, please consult the CPT coding manual that is published by The American Medical Association (AMA). If you have any questions regarding the use of a code, please contact your local Medicare carrier.

Specimen Requisition Form

Please PRINT all information. Complete a separate Test Requisition for each patient. Note: If more than one specimen is being submitted per patient, and specimens need to be stored and transported at different temperatures, it is your responsibility to use separate bags and Test Request forms for each different transport temperature.

Fill in all patient information (patient name, ID number, sex, date of birth, physician name, collection date and time, specimen type, diagnosis, ICD-10 code, physician identifiers, all physician information and forms must contain physician signature). Check the box indicating the appropriate test requested and submit original copy to laboratory along with the specimen (retain a copy for your records). Laboratory test orders can only be requested by and will only be performed for authorized persons. Physician electronic orders and or written physician orders for laboratory testing, may be attached to the lab request form and will be accepted in lieu of signed lab request form.

Additional lab request forms can be obtained via the web. If you do not have access, Clients may call us at (305)-243-5450. Please have your client account number available for faster service.

To establish a new client account number please call the Genetics Billing Office at (305)-243-6583. Communication may also be completed via email Geneticsbillingoffice@med.miami.edu. Please note that specimens will not be processed until billing functions have been approved and cleared.

Reference Lab Testing
Reference Lab Testing is NOT available Via the BMGDL. Please contact the BMGDL at (305)-243-5450 for more specific information.

**Supplies**
Biochemical and Molecular Genetics Diagnostic Laboratory does not provide any specimen collection supplies.

Should any question arise regarding acceptable specimen containers, clients may review the *Specimen Requirements* section of each test page and or refer to the BMDGL Test Tube Diagrams.

If additional information or assistance is required please contact the BMGDL laboratory performing the test assay at (305)-243-5450.

**Specimen Shipping Address**
Biochemical and Molecular Genetics Diagnostic Laboratory University of Miami, Miller School of Medicine

1501 NW 10 AVE  
BRB, Room 535- M860  
Miami, FL 33136  
[biocemgenlab@med.miami.edu](mailto:biocemgenlab@med.miami.edu)

**Letter Mailing Address:**
University of Miami, Miller School of Medicine  
HIHG  
PO Box 019132 (M860)  
Miami, FL 33101

**Out of State Specimens**
Clients should contact the laboratory prior to shipping any specimen from outside of the State of Florida. Please contact the BMGDL at (305)-243-5450.

Clients are requested to call the Genetics Billing Office at (305)-243-6583. Communication may also be completed via email: Geneticsbillingoffice@med.miami.edu. Please note that specimens will not be processed until out of State billing functions have been approved and cleared.

**Electronic Compendium**
An election version of this document is available at: [http://medgen.med.miami.edu/BMGDL/search-for-a-test](http://medgen.med.miami.edu/BMGDL/search-for-a-test). Click on Laboratory Compendium.
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*The Test List is provided on page 16*
### Quantitative Amino Acid Analysis, Plasma

Quantitative analysis of plasma amino acids and related compounds. Provides diagnostic information pertaining to certain amino acidopathies, organic acidemias, and other metabolic conditions.

**Test Methodology:** Anion Exchange Chromatography with colorimetric detection.

**Indications for Testing:** Many inborn errors of amino acid metabolism, (including phenylketonuria and tyrosinemia), may be identified. Amino acid disorders can manifest at any age, but most become evident in newborn infants or early childhood. Amino acid disorders result in the deficiency or accumulation of one or more amino acids (in biological fluids), which leads to the clinical signs and symptoms in the patient.

**Specimen Requirement(s):**
- 2.0 ml of whole blood (ideal)
- 1 ml plasma (ideal) OR
- 0.5 ml plasma (minimum)

**Collection Tube:** Green Top Tube /w heparin

**Handling:** Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma

**TAT:** Batch 8-10 business days

**CPT Code(s):** 82139

**Lab Code:** 110002

**Special Note:** Fasting is required for specimen collection. Send blood to lab on ice same day of collection. OR freeze plasma-transport frozen sample to lab within 14 days of sample collection.

**Fee:** Contact Billing Dept

### Organic Acid Analysis, Urine

Semi-quantitative analysis of excreted organic acids and related compounds. Provides diagnostic information regarding organic acidemias, fatty acid oxidation disorders, and other conditions.

**Test Methodology:**
This test is performed by using Solvent Extraction, gas chromatography, mass spectrometry.

**Indications for Testing:** The urine organic acids analysis provides a qualitative report of abnormal levels of organic acids via GC-MS.

Diagnostic specificity of inborn errors of metabolism via urine organic acids is variable due to factors such as specimen collection when the patient is asymptomatic versus acutely ill, taking dietary supplements, or anabolic versus catabolic.

**Specimen Requirement(s):**
- 10.0 ml of urine (ideal)
- 2.0 ml of urine (minimum)

**Collection Tube:** Urine container

**Handling:** Collect on ice and or freeze for transport

**TAT:** Batch 8-10 business days

**CPT Code(s):** 83919

**Lab Code:** 110013

**Special Note:** First morning void is preferred. Urine should be collected (without preservative) on ice and frozen. OR send fresh sample to lab on collection day ASAP. Transport frozen sample to lab within 14 days of sample collection.

**Fee:** Contact Billing Dept
**Acylcarnitine Analysis, Plasma**

Quantitative analysis of individual fatty acid-esterified carnitine species. Provides diagnostic information regarding fatty acid oxidation disorders and organic acidoses.

**Test Methodology:**
This test is performed by using stable isotope dilution via tandem mass spectrometry.

**Indications for Testing:**
The acylcarnitine analysis in plasma provides a diagnosis of fatty acid oxidation disorders and several organic acidurias.

This test is useful for evaluating treatment during follow-up of patients with fatty acid beta-oxidation disorders and several organic acidurias.

**Specimen Requirement(s):**
- 2.0 to 3.0 ml of whole blood (ideal)
- 0.5 ml whole blood (minimum)
- 1.0 ml plasma (ideal)
- 100 ul plasma (minimum)

**Collection Tube:** Green Top Tube /w heparin

**Handling:** Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma

**TAT:** Batch 8-10 business days

**CPT Code(s):** 82017

**Lab Code:** 11001

**Special Note:** Send blood to lab same day of collection. OR freeze plasma-transport frozen sample to lab within 14 days of sample collection.

**Fee:** Contact Billing Dept

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**Free/Esterified Carnitine Determination, Plasma**

Quantitative analysis of free and esterified carnitine fractions. Complementary to acylcarnitine analysis (see below); provides diagnostic information regarding fatty acid oxidation disorders and organic acidoses.

**Test Methodology:**
This test is performed by using stable isotope dilution via tandem mass spectrometry.

**Indications for Testing:**
Carnitine levels are disturbed in primary disorders of the carnitine cycle, or secondary disturbances of carnitine metabolism due to other biochemical disorders.

This test is useful for evaluation of patients with a clinical suspicion of a wide range of conditions including organic acidoses, fatty acid oxidation disorders, and primary carnitine deficiency.

**Specimen Requirement(s):**
- 1.0 to 2.0 ml of whole blood (ideal)
- 200 ul plasma (minimum)

**Collection Tube:** Green Top Tube /w heparin

**Handling:** Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma

**TAT:** Batch 8-10 business days

**CPT Code(s):** 82379

**Lab Code:** 110005

**Special Note:** Send blood to lab same day of collection ASAP. OR freeze
### MSUD Profile

**MSUD profile (Ala, Ile, Leu, Val)** Monitoring of patients with maple syrup urine disease

**Test Methodology:**
This test is performed by using anion exchange chromatography with colorimetric detection.

**Indications for Testing:**
Maple syrup urine disease (MSUD) is an inborn error of metabolism caused by the deficiency of the branched-chain ketoacid dehydrogenase (BCKDH) complex.

-Treatment of MSUD aims to normalize the concentration of branched chain amino acids (BCAA) by dietary restriction of these amino acids, and because BCAA belong to the essential amino acids, the dietary treatment requires frequent adjustment.

**Specimen Requirement(s):** 2.0 ml of whole blood (minimum)

<table>
<thead>
<tr>
<th>Collection Tube</th>
<th>Handling</th>
<th>TAT</th>
<th>CPT Code(s)</th>
<th>Lab Code</th>
<th>Special Note</th>
<th>Fee</th>
</tr>
</thead>
<tbody>
<tr>
<td>Green Top Tube /w heparin</td>
<td>Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma</td>
<td>Batch 8-10 business days</td>
<td>82136</td>
<td>110055</td>
<td>REFER TO AMINO ACIDS NOTES</td>
<td>Contact Billing Dept</td>
</tr>
</tbody>
</table>

### PKU Profile

**PKU profile (Phe, Tyr)** for the monitoring of affected patients

**Test Methodology:**
This test is performed by using anion exchange chromatography with colorimetric detection.

**Indications for Testing:**
Presymptomatic identification of disorders to allow for early initiation of treatment and consequent improvement in the long-term prognosis of affected patients

**Specimen Requirement(s):** 2.0 ml of whole blood (minimum)

<table>
<thead>
<tr>
<th>Collection Tube</th>
<th>Handling</th>
<th>TAT</th>
<th>CPT Code(s)</th>
<th>Lab Code</th>
<th>Special Note</th>
<th>Fee</th>
</tr>
</thead>
<tbody>
<tr>
<td>Green Top Tube /w heparin</td>
<td>Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma</td>
<td>Batch 8-10 business days</td>
<td>84030</td>
<td>110061</td>
<td>REFER TO AMINO ACIDS NOTES</td>
<td>Contact Billing Dept</td>
</tr>
</tbody>
</table>
Methylmalonic Acid Analysis, Plasma or Urine  TEST IS TEMPORARILY SUSPENDED

Quantitative analysis of excreted methylmalonic acid. Provides diagnostic information regarding disorders of methylmalonyl-CoA and vitamin B12 (cobalamin) metabolism

Test Methodology:
This test is performed by using stable isotope dilution via tandem mass spectrometry.

Indications for Testing:
This test is useful evaluating individuals with signs and symptoms associated with a variety of causes of cobalamin (vitamin B12) deficiency. May be used in the evaluation of children with signs and symptoms of methylmalonic acidemia.

Specimen Requirement(s):

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Volume Requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole Blood</td>
<td>2.0-3.0 ml (ideal)</td>
</tr>
<tr>
<td></td>
<td>0.5 ml (minimum)</td>
</tr>
<tr>
<td>Plasma</td>
<td>1.0 ml (ideal)</td>
</tr>
<tr>
<td></td>
<td>100 µl (minimum)</td>
</tr>
<tr>
<td>Urine</td>
<td>5.0 ml (ideal)</td>
</tr>
<tr>
<td></td>
<td>2.0 ml (minimum)</td>
</tr>
</tbody>
</table>

Collection Tube: Green Top Tube /w heparin OR Urine container

Handling: Refrigerate whole blood (4 degrees C, recommended). OR Freeze plasma OR Freeze urine

TAT: Batch 8-10 business days

CPT Code(s):
83921

Lab Code:
110046 Plasma
110047 Urine

Special Note:
Fasting is required for blood specimen collection. Send blood to lab same day of collection ASAP. First morning void is preferred. Urine should be collected on ice and frozen. Transport frozen sample to lab within 14 days of sample collection.

Fee: Contact Billing Dept
Succinylacetone Determination, Urine

Quantitative analysis of excreted succinylacetone. Provides diagnostic information regarding disorders of fumarylacetoacetic acid hydrolase (FAH).

**Test Methodology:**
This test is performed by using Solvent Extraction, gas chromatography, mass spectrometry.

**Indications for Testing:**
This test may be used to diagnose individuals with tyrosinemia type 1, in conjunction with organic and amino acids, or to monitor individuals with tyrosinemia type 1 who are on therapy.

**Specimen Requirement(s):**
- Handling: Collect on ice and freeze for transport
- TAT: Batch 8-10 business days
- CPT Code(s): 82542
- Lab Code: 110013

**Special Note:**
First morning void is preferred. Urine should be collected on ice and frozen. Transport frozen sample to lab within 14 days of sample collection.

**Fee:**
Contact Billing Dept
Galactosemia is a disorder of galactose metabolism that can result in life-threatening complications including feeding problems, failure to thrive, hepatocellular damage, bleeding, and sepsis in untreated infants. Data based on newborn screening (NBS) suggest a prevalence of 1:30,000 for classic galactosemia (G/G). If a lactose/galactose-restricted diet is provided during the first ten days of life, the neonatal symptoms quickly resolve and the complications of liver failure, sepsis, neonatal death, and intellectual disability can be prevented. Despite adequate treatment from an early age, children with galactosemia remain at increased risk for developmental delays, speech problems (termed "verbal dyspraxia"), and abnormalities of motor function. A female with galactosemia is at increased risk for premature ovarian insufficiency. Quantitative measurement of erythrocyte concentration of gal-1-P and erythrocyte GALT enzyme activity establishes the diagnosis of galactosemia. Molecular genetic testing is used to confirm the diagnosis of galactosemia and to distinguish the Duarte (D2) variant allele from the LA variant allele (ref GeneReviews Galactosemia).

**Test Methodology:**
This test is performed by using the Sanger’s method, which is also referred to as dideoxy sequencing or chain termination sequencing (Sanger et al., 1977). Briefly, genomic DNA segments of 100-500 nucleotides (in general containing the gene exons of interest plus an additional 20 base pairs from the exon/intron junction into the intron) are enriched by polymerase chain reaction (PCR). The products of this amplification reaction (amplicons) are then sequenced bi-directionally. In rare cases, the sequence can only be determined in one direction due to particular base pair sequences at the beginning of the extension product that makes the downstream sequence un-interpretable).

**Indications for Testing:**
- Individuals with a biochemical testing confirmed diagnosis of galactosemia or abnormal biochemical testing with neither or only one disease-causing mutation detected by targeted mutation analysis. In these individuals gene sequencing may help in identifying mutations not included in the targeted mutation analysis panel.
# TEST LIST

<table>
<thead>
<tr>
<th>Test Name</th>
<th>Effective Date</th>
<th>Primary Instrument</th>
<th>Methodology</th>
<th>Proficiency Testing Provider</th>
<th>Test Complexity (M=Moderate H=High)</th>
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<tbody>
<tr>
<td>Acylcarnitines</td>
<td>6-6-2016</td>
<td>Agilent 6460 LC-MS/MS (manufacturer Agilent)</td>
<td>Stable Isotope Dilution</td>
<td>CAP-BGL</td>
<td>High</td>
</tr>
<tr>
<td>Quantitative Amino Acids, Plasma</td>
<td>2-8-2016</td>
<td>Biochrom amino acid analyzer (manufacturer Biochrom)</td>
<td>Anion Exchange Chromatography with colorimetric detection</td>
<td>CAP-BGL ERNDIM (PAA)-APT</td>
<td>High</td>
</tr>
<tr>
<td>MSUD-Amino Acid Plasma</td>
<td>2-8-2016</td>
<td>Biochrom amino acid analyzer (manufacturer Biochrom)</td>
<td>Anion Exchange Chromatography with colorimetric detection</td>
<td>ERNDIM (PAA)-APT</td>
<td>High</td>
</tr>
<tr>
<td>Organic Acids, Urine</td>
<td>2-8-2016</td>
<td>Agilent 6890N/5975 GC-MS/MS (manufacturer Agilent)</td>
<td>Solvent Extraction, gas chromatography, mass spectrometry</td>
<td>CAP-BGL ERNDIM (QOA)-APT</td>
<td>High</td>
</tr>
<tr>
<td>PKU-Amino Acid Plasma</td>
<td>2-8-2016</td>
<td>Biochrom amino acid analyzer (manufacturer Biochrom)</td>
<td>Anion Exchange Chromatography with colorimetric detection</td>
<td>ERNDIM (PAA)-APT</td>
<td>High</td>
</tr>
<tr>
<td>Total &amp; Free Carnitine</td>
<td>7-8-2016</td>
<td>Agilent 6460 LC-MS/MS (manufacturer Agilent)</td>
<td>Stable Isotope Dilution</td>
<td>CAP-BGL-1 ERNDIM (SAS)-APT</td>
<td>High</td>
</tr>
<tr>
<td>Succinylacetone, Urine</td>
<td>9-9-2016</td>
<td>Agilent 6890N/5975 GC-MS/MS (manufacturer Agilent)</td>
<td>Solvent Extraction, gas chromatography, mass spectrometry</td>
<td>ERNDIM (SAU)-APT</td>
<td>High</td>
</tr>
<tr>
<td>Methylmalonic Acid, Plasma</td>
<td>3-16-2017</td>
<td>Agilent 6460 LC-MS/MS (manufacturer Agilent)</td>
<td>Stable Isotope Dilution</td>
<td>ERNDIM (SAS)-APT</td>
<td>High</td>
</tr>
<tr>
<td>Methylmalonic Acid, Urine</td>
<td>3-16-2017</td>
<td>Agilent 6460 LC-MS/MS (manufacturer Agilent)</td>
<td>Stable Isotope Dilution</td>
<td>ERNDIM (SAU)-APT</td>
<td>High</td>
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**METHYLMALONIC ACID, TEST TEMPORARILY SUSPENDED**

**METHYLMALONIC ACID, TEST TEMPORARILY SUSPENDED**
Biochemical and Molecular Genetics Diagnostic Laboratory
University of Miami, Miller School of Medicine

|----------------------------|--------|------------------------------------------|---------------|------------------|------|

References


Howanitz, J. and Howanitz, P, *Laboratory Results*; American Society of Clinical Pathologists 2001;116:311-315

Mayo Medical Labs Compendium; https://www.mayomedicallaboratories.com/test-catalog/Specimen