

INSTITUTE OF METABOLIC DISEASE
 BAYLOR RESEARCH INSTITUTE
 3812 ELM STREET
 DALLAS, TX 75226
 PHONE: (214) 820 - 4533 FAX: (214) 820 - 4853
 WEBSITE: www.baylorhealth.edu/imd

MASS SPECTROMETRY LABORATORY
 Director – Larry Sweetman, Ph.D.
 E-Mail: larrys@baylorhealth.edu
 Asst Director – Xiaowei Fu, M.D., Ph.D.
 E-Mail: xiaoweif@baylorhealth.edu

MASS SPECTROMETRY CLINICAL TESTS FOR NEW YORK

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
ACYLCARNITINE PROFILE * <i>See notes on preferred specimen types below</i>	1. Dried Blood Spot card (preferred) – 3 completely filled spots 2. Plasma - 0.2 mL of heparinized plasma Minimum volume – 0.1mL 3. Whole Blood – 3 mL of whole blood collected in a heparin tube Minimum volume – 2 mL 4. Serum – 0.2mL of serum Minimum volume – 0.1mL	1. Dried Blood spot – ship at room temperature. 2. Plasma or Serum – Freeze plasma and ship overnight on 3-4 lbs. of dry ice. 3. Whole Blood – ship at room temperature. Ship all sample types using a guaranteed overnight courier.	1 - 3 working days from receipt of sample <i>(70% complete in 1 day)</i>	82017	\$ 110.00
CARNITINE LEVELS <i>See notes on preferred specimen types below</i>	1. Dried Blood Spot card – 3 completely filled spots 2. Plasma (preferred) - 0.2 mL of heparinized plasma. Minimum volume – 0.1mL 3. Serum – 0.2mL of serum Minimum volume – 0.1mL	1. Dried Blood spot – ship at room temperature. 2. Plasma or Serum – Freeze plasma and ship overnight on 3-4 lbs. of dry ice. Ship all sample types using a guaranteed overnight courier.	1 - 3 working days from receipt of sample <i>(75% complete in 1 day)</i>	82379	\$ 80.00
ORGANIC ACIDS (Quantitative GC/MS)	Urine – optimal sample volume is 5 mL Minimum sample volume – 2 mL Collect urine in a sterile container with no preservatives	Freeze urine and ship overnight on 3-4 lbs of dry ice using a guaranteed overnight courier.	3 - 7 working days from receipt of sample <i>(75% complete in 4 days)</i>	83918	\$ 195.00

* **PRENATAL DIAGNOSIS** of Propionic Acidemia and Methylmalonic Acidemia is available by this method. Detection of additional disorders may also be available. Contact the Laboratory Director to discuss and to obtain all other pertinent information (Sample Requirements, Shipping and Handling, Turnaround Time, CPT Code and Cost). **APPROVAL FROM THE LABORATORY DIRECTOR MUST BE OBTAINED PRIOR TO SENDING SAMPLES FOR PRENATAL DIAGNOSIS.**

NOTES ON PREFERRED SPECIMEN TYPES

ACYLCARNITINE PROFILE – The preferred specimen is **dried blood spots** because the long-chain acylcarnitines are absorbed on the surface of the red cells so that the normal levels are much higher for dried blood spots than in plasma. Therefore the elevations of these in some milder forms of long-chain fatty acid oxidation disorders may not be as reliably detected in the plasma as they are in the dried blood spots. Serum specimens are acceptable.

CARNITINE LEVELS – The preferred sample is **plasma** because the free carnitine levels in plasma reflect the circulating available free carnitine and physicians are more familiar with the normal ranges for plasma free carnitine. Serum specimens are acceptable. The reference ranges for dried blood spot free carnitine is lower than for plasma due to lower levels in the red cells.

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MOLECULAR DIAGNOSTIC LABORATORY
 Director – Bingzhi Yang, M.D.
 E-mail: bingzhiy@baylorhealth.edu

MOLECULAR DIAGNOSTICS CLINICAL TESTS FOR NEW YORK

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
LCHAD DNA * (1528 G>C)	1. Dried blood spot card (preferred) – 5 completely filled spots 2. Whole Blood –1 mL collected in an EDTA (lavender top) tube or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. Minimum sample volume – 0.5 mL 3. Fibroblasts - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2 weeks from receipt of sample	83890 83892 83894 83898 83912	\$ 150.00
MCAD DNA * (985 A>G)	1. Dried blood spot card (preferred) – 5 completely filled spots 2. Whole Blood - 1 mL collected in an EDTA (lavender top) or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. Minimum sample volume – 0.5 mL 3. Fibroblasts - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2 weeks from receipt of sample	83890 83892 83894 83898 83912	\$ 150.00

PRENATAL DIAGNOSIS is available by this method. Contact the IMD Medical Director, the Molecular Diagnostics Laboratory Director or the Mass Spectrometry Laboratory Director to discuss and to obtain all other pertinent information (Sample Requirements, Shipping and Handling, Turnaround Time, CPT Code and Cost).
APPROVAL FROM ONE OF THE DIRECTORS MUST BE OBTAINED PRIOR TO SENDING SAMPLES FOR PRENATAL DIAGNOSIS.

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TISSUE CULTURE LABORATORY
 Medical Director – Charles R. Roe, M.D.
 Email: charlesr@baylorhealth.edu
 Lab Director – Larry Sweetman, Ph.D.
 E-Mail: larrys@baylorhealth.edu

TISSUE CULTURE CLINICAL TESTS FOR NEW YORK

TEST NAME	AMOUNT OF SAMPLE	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
MITOCHONDRIAL BETA OXIDATION DEFECTS * *	<ol style="list-style-type: none"> Fibroblasts – 2 sub-confluent T-25 flasks filled with medium supplemented with serum. Amniocytes – **This RESEARCH prenatal test must be approved by the IMD Medical Director or the Mass Spectrometry Laboratory Director PRIOR to sending the cultured amniocytes. Please contact the IMD Medical Director or the Mass Spectrometry Laboratory Director for information. 	<p>Shipping:</p> <ol style="list-style-type: none"> Send all sample types at room temperature. Package samples carefully to avoid breaking or freezing. Using indelible ink, flasks must be labeled with the patient's name, culture date and passage number. Samples should be shipped using a guaranteed overnight courier. Ship samples only Monday through Wednesday to ensure receipt. <p>Handling:</p> <ol style="list-style-type: none"> DO NOT SEND MYCOPLASMA POSTIVE CELLS. Cell lines known to be positive for mycoplasma will not be accepted. Wrap the neck of the flask with parafilm or foil to prevent contamination and leakage. 	2 – 4 weeks from receipt of sample <i>(80 % completed within 2 – 3 weeks)</i>	88233 84157 83789	\$ 750.00

* * Approval of this prenatal research testing request must be obtained from the IMD Medical Director or the Mass Spectrometry Laboratory Director prior to sending cultured amniocytes. Gestational age-matched controls ARE REQUIRED for accurate interpretation of these results.

INSTITUTE OF METABOLIC DISEASE
3812 Elm Street
Dallas, TX 75226

Phone: (214) 820 – 4533
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Patient Name: _____

D.O.B./Age: _____ Sex: MALE FEMALE

Medical Record # or Patient ID #: _____

NEW YORK CLINICAL TEST REQUISITION FORM

Specimen Information – Sample 1

Accession/Lab ID # _____
Specimen Type: _____
Sample Date: _____ Time: _____
For Cell Lines- Flask Date: _____
Passage #: _____

Specimen Information – Sample 2

Accession/Lab ID # _____
Specimen Type: _____
Sample Date: _____ Time: _____
For Cell Lines- Flask Date: _____
Passage #: _____

Ordering Physician: _____

Phone: _____ Fax: _____

NY State Clientele MUST check one of the following: Informed consent form for Genetic Testing enclosed, Informed consent form for Genetic Testing on file in Physician's office, Physician has signed or initialed above indicating that information regarding the nature of the Genetic Testing was conveyed to the patient.

TO ASSIST INTERPRETATION PLEASE FILL IN BELOW

Primary Presenting Symptoms: _____

Abnormal Labs: _____

Suspected Diagnosis: _____

Diet or Infant Formula: _____

Medications: _____

(If this space is not sufficient please attach clinical summary or patient history)

TEST(S) REQUESTED

(Please check all that apply for Sample 1)

Mass Spectrometry tests:

- Acylcarnitine Profile*
- Carnitine Levels*
- Organic Acids*

Molecular Diagnostic tests:

- LCHAD DNA (1528G>C)
- MCAD DNA analysis(985A>G)

Tissue Culture tests:

- Mitochondrial Beta Oxidation Defects

* **Please Note:** For age related reference ranges, Date of Birth and Sample Date is required with each request.

TEST(S) REQUESTED

(Please check all that apply for Sample 2)

Mass Spectrometry tests:

- Acylcarnitine Profile*
- Carnitine Levels*
- Organic Acids*

Molecular Diagnostic tests:

- LCHAD DNA (1528G>C)
- MCAD DNA analysis(985A>G)

Tissue Culture tests:

- Mitochondrial Beta Oxidation Defects

* **Please Note:** For age related reference ranges, Date of Birth and Sample Date is required with each request.

RESULTS ADDRESS

Phone: _____ Fax: _____

BILLING ADDRESS (The IMD does not bill patients, Medicare, Medicaid or insurance)

Phone: _____ Fax: _____

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Medical Director – Charles R. Roe, M.D.

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ADDITIONAL LABORATORY AND SHIPPING INFORMATION

LABORATORY HOURS: MONDAY - FRIDAY 8:30 a.m. – 5:00 p.m. (C.S.T.)

Shipping Information:

- Please use only guaranteed overnight couriers (FedEx, DHL, and UPS) to insure NEXT DAY TRACKABLE delivery.
- As per CLIA and CAP regulations, all specimens must be submitted with a completed test requisition.
- **Samples submitted from New York must have a completed informed consent form or must indicate on the test requisition that consent has been obtained.**
- All specimens must be labeled with the patient name and sample collection date.
- Use indelible ink or gummed labels to label samples.
- Place samples inside a specimen transport bag and the associated documents inside the pouch in the specimen transport bag. Do not place the documentation inside the specimen transport bag with the sample.
- Ship samples **Monday through Thursday only. Please note that samples for Mitochondrial Beta Oxidation Defects must be shipped Monday through Wednesday only. NO Saturday deliveries will be accepted.**

Testing:

- For all PRENATAL tests, approval MUST be obtained from the IMD Medical Director or the appropriate laboratory director PRIOR to sample submission.
- For STAT analysis, please contact the appropriate laboratory director to provide clinical information. STAT analyses will not be performed unless the clinical information is provided.
- STAT analyses will not be performed on Supplemental Newborn Screening samples.

Result Reporting:

- **Only CRITICAL results are reported immediately by telephone and fax.**
- Results are available for a VERBAL report (or if possible, a preliminary fax on request) within the turnaround time specified for each test.
- Result reports are faxed and mailed to the submitter and physician (if physician information is provided).

Billing:

- **The IMD does not bill patients, Medicare, Medicaid or insurance.** Please contact our Billing department at (214) 820-4533 with questions about test prices, CPT codes, billing or invoicing.