

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

MASS SPECTROMETRY LABORATORY
 Director – Larry Sweetman, Ph.D.
 E-Mail: larrys@baylorhealth.edu

MASS SPECTROMETRY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
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 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
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 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

* **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.

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

MASS SPECTROMETRY LABORATORY
 Director – Larry Sweetman, Ph.D.
 E-Mail: larrys@baylorhealth.edu

MASS SPECTROMETRY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
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
EVEROLIMUS/ ZORTRESS	Whole blood (EDTA) lavender top – optimum volume - 3mL, 0.5mL minimum. Routine phlebotomy. Trough levels should be obtained: 12-18 hours after oral dose, 12 hours after intravenous dose, or immediately prior to next dose	Handling: Freeze and store at -20°C until shipped Shipping: Ship sample overnight with 3-4 lbs of dry ice using a guaranteed overnight courier.	Same day turnaround if received by 9 AM	80299
SIROLIMUS/ RAPAMYCIN	Whole blood (EDTA) lavender top – optimum volume - 3mL, 0.5mL minimum. Routine phlebotomy. Trough levels should be obtained: 12-18 hours after oral dose, 12 hours after intravenous dose, or immediately prior to next dose	Handling: Freeze and store at -20°C until shipped Shipping: Ship sample overnight with 3-4 lbs of dry ice using a guaranteed overnight courier.	Same day turnaround if received by 9 AM	80195

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

NEUROPHARMACOLOGY LABORATORY
 Director – Teodoro Bottiglieri, Ph.D.
 E-mail: teodorob@baylorhealth.edu

NEUROPHARMACOLOGY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
AMINO ACIDS	<p>Plasma (preferred) – 0.5 mL of heparinized plasma</p> <p>Serum – 0.5 mL of serum collected in a red-top tube</p> <p>CSF – 0.5 mL collected in a sterile container and frozen at -80°C</p> <p>Absolute minimum for all sample types – 150 µL</p>	For all sample types, freeze and ship overnight on 3-4 lbs of dry ice. Ship using a guaranteed overnight courier.	1 week from sample receipt	82139
GABA – Free and Total	<p>CSF – 0.5 mL collected in a sterile container and frozen at -80°C; minimum volume – 250µL.</p> <p>SPECIAL NOTE: Can be performed using Tube #5 from Special Collection tubes/kit – separate collection not necessary</p>	Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from sample receipt	83789
HOMOCYSTEINE (total)	<p>Plasma – 0.5 mL of heparinized plasma</p> <p>Serum – 0.5 mL</p>	For both sample types, centrifuge the sample within 1 hour of collection. Freeze sample and ship overnight on 3-4 lbs of dry ice. Ship using a guaranteed overnight courier.	1 week from sample receipt	83090
5-METHYLTETRAHYDROFOLATE (5-MTHF) in PLASMA or SERUM	<p>Plasma – 0.5 mL of heparinized plasma</p> <p>Serum – 0.5 mL</p>	For both sample types, centrifuge the sample within 1 hour of collection. For all sample types, freeze and ship overnight on 3-4 lbs of dry ice. Ship using a guaranteed overnight courier.	1 week from sample receipt	83789
LACTATE - <i>Cerebrospinal Fluid</i>	0.5 ml - CSF; frozen at -80°C at collection.	Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from sample receipt.	83605

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

NEUROPHARMACOLOGY LABORATORY
 Director – Teodoro Bottiglieri, Ph.D.
 E-mail: teodorob@baylorhealth.edu

NEUROPHARMACOLOGY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
5-METHYLTETRAHYDROFOLATE (5-MTHF) - Cerebrospinal Fluid See Special Note below	0.5 ml – CSF; frozen at -80°C at collection. <u>See next sheet for CSF collection instructions</u>	Call lab for SPECIAL Collection tubes and kit Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82491
MONOAMINE NEUROTRANSMITTER METABOLITES (5-HIAA, HVA, 3-OMD) Cerebrospinal Fluid	0.5 ml – CSF; frozen at -80°C at collection. <u>See next sheet for CSF collection instructions</u>	Call lab for SPECIAL Collection tubes and kit Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82492
TETRAHYDROBIOPTERIN & NEOPTERIN PROFILE (BH4,N) Cerebrospinal Fluid	0.5 ml - CSF; frozen at -80°C at collection. <u>See next sheet for CSF collection instructions</u>	Call lab for SPECIAL Collection tubes and kit Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82492
NEOPTERIN - Cerebrospinal Fluid See Special Note below	0.5 ml - CSF; frozen at -80°C at collection. <u>See next sheet for CSF collection instructions</u>	Call lab for SPECIAL Collection tubes and kit Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82491

SPECIAL NOTE: Both 5-Methyltetrahydrofolate and Neopterin can also be run from a single tube collection of 0.5 mL of CSF; it is not necessary to collect a whole kit if you are only requesting these two tests!

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

NEUROPHARMACOLOGY LABORATORY
 Director – Teodoro Bottiglieri, Ph.D.
 E-mail: teodorob@baylorhealth.edu

NEUROPHARMACOLOGY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
Methylenetetrahydrofolate Reductase C677T MTHFR C677T	<ol style="list-style-type: none"> Whole Blood spotted on FTA card (preferred) – see special collection instructions Whole blood – collect 1-2 mL of whole blood in a vacutainer tube containing EDTA, citrate, or heparin-based anticoagulants Purified Genomic DNA – <ol style="list-style-type: none"> Sample volume: 20 to 40 uL Required concentration: 1 to 20 ng 	<p>HANDLING:</p> <ol style="list-style-type: none"> Whole blood – store at -20°C for up to 1 year. Whole blood specimens spotted on FTA cards are stable for up to 17 years stored at room temperature. Genomic DNA – store at 4°C until shipment <p>SHIPPING:</p> <ol style="list-style-type: none"> Whole Blood and Genomic DNA samples – ship with a cold pack using a trackable courier. FTA cards - ship at room temperature using a trackable courier. 	7 days from sample receipt	81291
Methylenetetrahydrofolate Reductase A1298C MTHFR A1298C	<ol style="list-style-type: none"> Whole Blood spotted on FTA card (preferred) – see special collection instructions Whole blood – collect 1-2 mL of whole blood in a vacutainer tube containing EDTA, citrate, or heparin-based anticoagulants Purified Genomic DNA – <ol style="list-style-type: none"> Sample volume: 20 to 40 uL Required concentration: 1 to 20 ng 	<p>HANDLING:</p> <ol style="list-style-type: none"> Whole blood – store at -20°C for up to 1 year. Whole blood specimens spotted on FTA cards are stable for up to 17 years stored at room temperature. Genomic DNA – store at 4°C until shipment <p>SHIPPING:</p> <ol style="list-style-type: none"> Whole Blood and Genomic DNA samples – ship with a cold pack using a trackable courier. FTA cards - ship at room temperature using a trackable courier. 	7 days from sample receipt	81291

INSTITUTE OF METABOLIC DISEASE
BAYLOR RESEARCH INSTITUTE
3812 ELM STREET
DALLAS, TX 75226

PHONE: (214) 820 - 4533 FAX: (214) 820 – 4853

WEBSITE: <http://www.baylorhealth.edu/IMD>

NEUROPHARMACOLOGY LABORATORY
Director – Teodoro Bottiglieri, Ph.D.
E-mail: teodorob@baylorhealth.edu

CSF COLLECTION PROTOCOL:

CSF Collection Protocol for the measurement of Monoamine Neurotransmitter Metabolites, Tetrahydrobiopterin and Neopterin Profile or 5-Methyltetrahydrofolate metabolite assays (these specimens may be used for lactate and amino acids as well, if requested)

1. **THE CSF MUST BE COLLECTED IN OUR SAMPLE COLLECTION TUBES.** Call our laboratory to obtain appropriate sample collection tubes. Each sample collection set consists of 5 microcentrifuge tubes in a cardboard holder. Tube #3 contains antioxidants necessary to protect the sample integrity. **One set of tubes is required per patient.**
2. **CSF MUST BE collected from the first drop into the designated tubes in the order indicated in the following table.** Fill each tube to the marked line with the following volumes:

Tube Number	Required volume	The total CSF volume required is 3.5 ml
1	0.5 mL	
2	0.5 mL	
3	1.0 mL	
4	1.0 mL	
5	0.5 mL	

FAILURE TO FOLLOW THE COLLECTION INSTRUCTIONS MAY RESULT IN SAMPLE REJECTION.

DO NOT COLLECT THE CSF IN ONE LARGE TUBE AND ALIQUOT INTO THE TUBE SET!

- If the samples are not blood contaminated, place the tubes on ice (or dry ice if available) at the bedside. Transfer the samples to a -80°C freezer ASAP.
- **If the sample is blood contaminated, the tubes should immediately be centrifuged (prior to freezing) and the clear CSF transferred to new similarly labeled tubes then frozen and stored at -80°C ASAP. BLOOD CONTAMINATED SAMPLES WILL BE REJECTED!**
- Store all samples at -80°C until transport.

Please contact the Neuropharmacology Laboratory at 214-820-4533 if you have any additional questions or need to request the special sample collection tubes.

FOR ALL TEST REQUESTS:

1. Complete the test requisition. Test requisition forms are included in the IMD sample collection package or may be downloaded from our website, <http://www.baylorhealth.edu/Research/InstitutesCenters/IMD>. Please include on the requisition: tests required, sample date, date of birth, current medications and relevant history.
2. Verify that the samples are labeled properly with the Patient Name (first and last) and ID number.

SHIPPING:

1. 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 samples.
2. Ship the samples on dry ice using an overnight courier to the address above. Please use only guaranteed overnight couriers (FedEx, DHL, UPS) to insure Next Day delivery. **Ship Monday –Thursday ONLY.**

INSTITUTE OF METABOLIC DISEASE
BAYLOR RESEARCH INSTITUTE
3812 ELM STREET
DALLAS, TX 75226

PHONE: (214) 820 - 4533 FAX: (214) 820 – 4853

WEBSITE: <http://www.baylorhealth.edu/IMD>

NEUROPHARMACOLOGY LABORATORY
Director – Teodoro Bottiglieri, Ph.D.
E-mail: teodorob@baylorhealth.edu

BLOOD COLLECTION PROTOCOL: 1. BLOOD FROM FINGER STICK SPOTTED ON FTA CARD; 2. WHOLE BLOOD

(Preferred specimens are blood spotted on FTA Whatman card. Blood spotted on other newborn screening cards such as Whatman 903 cards are acceptable.)

1. FTA WHATMAN CARD BLOOD COLLECTION

- Patient must be lying down or seated during the entire procedure.
- The non-dominant hand is preferred, positioned below the heart.
- The 3rd and 4th fingers on the plantar side are the sites of choice.
- Cleanse the finger tip with alcohol prep. Allow to dry.
- Using a sterile lancet device, puncture skin just off center of the finger pad.
- Gently massage the patient's finger to force blood to the tip.
- Apply pressure to the side of the finger (avoid excessive pressure).
- Drip blood in the center of the circle and allow it to diffuse out. Only one circle is required.
- When collection has finished have the patient hold sterile gauze onto puncture site until bleeding stops.
- Allow blood to dry on card for 10 minutes.
- **Important:** Place labeled blood spot card in card holder and label with patients name, DOB and date of collection.

Please contact the Neuropharmacology Laboratory at 214-820-4533 if you have any additional questions or need to request the special FTA blood spot collection kits.

2. WHOLE BLOOD COLLECTION PROTOCOL

- Specimens are whole blood collected in a vacutainer tube containing EDTA, citrate, or heparin-based anticoagulants. *(Minimum volume required is 1.0 mL)*

FOR ALL TEST REQUESTS:

1. Complete the test requisition. Test requisition forms are included in the IMD sample collection package or may be downloaded from our website, www.baylorhealth.edu/imd. Please include on the requisition: tests required, sample date, date of birth, current medications and relevant history.
2. Verify that the samples are labeled properly with the Patient Name (first and last) and ID number.

SHIPPING:

1. Return FTA blood spot cards using a trackable service.
2. Ship whole blood samples on cool packs using an overnight courier to the address above. Please use only guaranteed couriers (FedEx, DHL, UPS) to insure delivery. Overnight shipping is recommended. **Ship Monday –Thursday ONLY.**

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

TISSUE CULTURE LABORATORY
 Lab Director – Larry Sweetman, Ph.D.
 E-Mail: larrys@baylorhealth.edu

TISSUE CULTURE CLINICAL TESTS

TEST NAME	AMOUNT OF SAMPLE	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE
MITOCHONDRIAL BETA OXIDATION DEFECTS	Fibroblasts – 2 sub-confluent T-25 flasks filled with medium supplemented with serum.	<p>Shipping:</p> <ol style="list-style-type: none"> 1. Send all sample types at room temperature. Package samples carefully to avoid breaking or freezing. 2. Using indelible ink, flasks must be labeled with the patient's name, culture date and passage number. 3. 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"> 1. DO NOT SEND MYCOPLASMA POSTIVE CELLS. Cell lines known to be positive for mycoplasma will not be accepted. 2. Wrap the neck of the flask with parafilm or foil to prevent contamination and leakage. 	4-6 weeks from receipt of sample	84157 83789

INSTITUTE OF METABOLIC DISEASE
3812 Elm Street
Dallas, TX 75226

Phone: (214) 820 – 4533
Fax: (214) 820 – 4853

MASS SPECTROMETRY AND TISSUE CULTURE
CLINICAL TEST REQUISITION FORM

Patient Name: _____
D.O.B./Age: _____ Gender: MALE FEMALE
Medical Record # or Patient ID #: _____

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. A Waiver from NY State has been obtained for the testing.

TEST(S) REQUESTED
(Please check all that apply for Sample 1)

TEST(S) REQUESTED
(Please check all that apply for Sample 2)

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)

- Mass Spectrometry tests:**
- Acylcarnitine Profile *
 - Carnitine Levels *
 - Organic Acids *
 - Everolimus/Zortress
 - Sirolimus/Rapamycin

- Mass Spectrometry tests:**
- Acylcarnitine Profile *
 - Carnitine Levels *
 - Organic Acids *
 - Everolimus/Zortress
 - Sirolimus/Rapamycin

- Tissue Culture tests:**
- Mitochondrial Beta Oxidation Defects *

- 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.

*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: _____

INSTITUTE OF METABOLIC DISEASE
3812 Elm Street
Dallas, TX 75226

Phone: (214) 820 – 4533
Fax: (214) 820 – 4853

Patient Name: _____
D.O.B./Age: _____ Gender: MALE FEMALE
Medical Record # or Patient ID #: _____

NEUROPHARMACOLOGY CLINICAL TEST REQUISITION FORM

Specimen Information

Accession/Lab ID # _____
Specimen Type: _____
Sample Date: _____ Time: _____

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. A Waiver from NY State has been obtained for the testing.

TEST(S) REQUESTED

- Amino Acids*
- GABA – Free and Total *
- Homocysteine (total)* - SPECIAL NOTE: Total Homocysteine in CSF is a research test
- Lactate
- Monoamine Neurotransmitter Metabolites*
- Tetrahydrobiopterin and Neopterin*
- Neopterin *
- 5-Methyltetrahydrofolate (5-MTHF) *

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

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)

RESULTS ADDRESS

Phone: _____ Fax: _____

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

Phone: _____ Fax: _____

IMPORTANT SPECIMEN COLLECTION INFORMATION FOR CSF SAMPLES

1. CSF must be collected using the special collection tube set according to the instructions detailed on the CSF Collection Protocol. Failure to collect the sample according to this protocol could result in sample rejection.
2. **DO NOT COLLECT THE CSF SAMPLE IN ONE LARGE TUBE THEN ALIQUOT INTO THE TUBE SET!**
3. **BLOOD CONTAMINATED SAMPLES WILL BE REJECTED!**
4. For more information and to request CSF Collection tube sets, contact the laboratory at 214-820-4533 or visit our website, www.baylorhealth.edu/IMD .

INSTITUTE OF METABOLIC DISEASE

3812 Elm Street
Dallas, TX 75226

Phone: (214) 820 – 4533
Fax: (214) 820 – 4853

NEUROPHARMACOLOGY CLINICAL TEST REQUISITION FORM

Specimen Information

Patient Name: _____
D.O.B./Age: _____ Gender: MALE FEMALE
Medical Record # or Patient ID #: _____

Accession/Lab ID # _____
Specimen Type: _____
Sample Date: _____ Time: _____

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. A Waiver from NY State has been obtained for the testing.

TEST(S) REQUESTED

- Methylene tetrahydrofolate Reductase (MTHFR) C677T
- Methylene tetrahydrofolate Reductase (MTHFR) A1298C

***Please Note:** Ensure that the FTA card and the multi-barrier pouch have required patient identification information.

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)

RESULTS ADDRESS

Phone: _____ Fax: _____

IMPORTANT SPECIMEN COLLECTION INFORMATION FOR FTA BLOOD SPOT KITS

Follow instructions for sample collection using the FTA blood spot kits provided. Inadequate collections and improperly labeled cards will result in rejection of specimens and the client will be notified.

For more information and to request FTA blood spot collection kits, contact the laboratory at 214-820-4533 or visit our website, <http://www.baylorhealth.edu/IMD>

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

Phone: _____ Fax: _____

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

INSTITUTE OF METABOLIC DISEASE
Medical Director – Dr. Raphael Schiffmann, M.D.,
M.H.Sc

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.