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

MASS SPECTROMETRY CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
ACYLCARNITINE PROFILE * *See notes on preferred specimen types below*	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 *(70% complete in 1 day)*	82017	$110.00
CARNITINE LEVELS *See notes on preferred specimen types below*	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 *(75% complete in 1 day)*	82379	$80.00

* **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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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 *(75% complete in 4 days)*	83918	**$195.00**
FABRY DISEASE SCREENING Gb3 (CTH, GL-3) Isoforms in Urine Cards – Quantitative	**Liquid Urine:** Urine – optimal sample volume is 10 mL; Minimum sample volume – 3 mL **IMPORTANT NOTES**: Collect the first morning void in a clean, sterile container with no preservatives. Must be a clean-catch sample. If frozen urine is received by the IMD, 3 urine cards will be prepared in our laboratory. **Urine spotted on filter paper:** Requires a special sample collection kit! Please contact the IMD at 214-820-4533 to request a kit and instructions for sample collection; the kit is available at no charge.	**For Liquid Urine:** Freeze urine and ship overnight on 3-4 lbs of dry ice using a guaranteed overnight courier. **For Urine spotted on filter paper:** Place the dried squares in the glassine envelope provided in the IMD collection kit. Place the glassine envelope and the completed test requisition inside the provided prepaid envelope and mail the sample. For trackable shipments, please use FedEx or DHL.	7-10 working days from sample receipt	83789	**$50.00**

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

MOLECULAR DIAGNOSTICS CLINICAL TESTS

TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
ALPHA-1 ANTITRYPSIN DEFICIENCY – COMMON MUTATION (E342K) ANALYSIS * OR INDIVIDUAL KNOWN SERPINA1 MUTATION (NOTE –OTHER THAN THE MUTATIONS LISTED IN THE PANEL BELOW)	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 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 83891 83892 83894 83898 83904 83909 83912	**$150.00 for common mutation** **$250.00 for other mutations**
ALPHA-1 ANTITRYPSIN DEFICIENCY – PANEL OF 8 MUTATIONS OR INDIVIDUAL MUTATION • Glu342Lys • Val361Ala • Leu362Met • Gly373Trp • Thr416Pro • Pro386 Ser • Glu400Asp • Gln401Lys • MAY ORDER INDIVIDUAL MUTATIONS LISTED ON PANEL	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-6 weeks from receipt of sample	83890 83891 83892 83894 83898 83904 83909 83912	Panel Cost- $300.00 **Individual mutation cost - $150.00**
ALPHA-1 ANTITRYPSIN DEFICIENCY – SERIPINA1 MUTATION ANALYSIS BY DNA SEQUENCING	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-8 weeks from sample receipt	83890 83891 x 6 83892 83894 x 6 83898 x 6 83904 x 6 83909 x 6 83912	**$600.00**

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
EIF2B -RELATED DISORDER - CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINATION/VANISHING WHITE MATTER DISEASE (CACH/VWM) MUTATION ANALYSIS USING DNA SEQUENCING INCLUDES TESTING OF: 1. EIF2B1 (ALPHA SUBUNIT) 2. EIF2B2 (BETA SUBUNIT) 3. EIF2B3 GAMMA SUBUNIT) 4. EIF2B4 (DELTA SUBUNIT) 5. EIF2B5 (EPSILON SUBUNIT)	1. **Whole Blood (preferred)** - 6 mL of whole blood collected in an EDTA (lavender top) or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. Minimum sample volume- 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	8-12 weeks from receipt of sample	83890 83891x58 83892 83894x58 83898x58 83904x58 83909x58 83912	$5000.00
EIF2B -RELATED DISORDER - CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINATION/VANISHING WHITE MATTER DISEASE (CACH/VWM) MUTATION ANALYSIS OF INDIVIDUAL EIF2B SUBUNIT GENES – SPECIFY SUBUNIT ON TEST REQUISITION	1. **Whole Blood (preferred)** - 6 mL of whole blood collected in an EDTA (lavender top) or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. **Minimum sample volume-** 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-6 weeks from receipt of sample	Based upon the subunit – please contact IMD Business Office	EIF2B1 - $950.00 EIF2B2 – $850.00 EIF2B3 - $1150.00 EIF2B4 - $1250.00 EIF2B5 – $1350.00

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
EIF2B -RELATED DISORDER - CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINATION/VANISHING WHITE MATTER DISEASE (CACH/VWM) MUTATION ANALYSIS FOR COMMON MUTATION R113H + PANEL OF 50 KNOWN MUTATIONS	1. **Whole Blood (preferred)** - 6 mL of whole blood collected in an EDTA (lavender top) or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. Minimum sample volume- 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-6 weeks from receipt of sample	83890 83891 83892 83894 83898 83904 83909 83912	$850.00
EIF2B -RELATED CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINATION/VANISHING WHITE MATTER(CACH/VWM) TARGET OR CARRIER MUTATION ANALYSIS *	1. **Whole Blood (preferred)** - 6 mL of whole blood collected in an EDTA (lavender top) or ACD (yellow top) tube. For ACD tube, either solution A or B is acceptable. Minimum sample volume- 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2-4 weeks from receipt of sample	83890 83891 83892 83894 83898 83904 83909 83912	$250.00

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CPT II DNA ANALYSIS– COMMON MUTATION (S113L) * OR OTHER INDIVIDUAL KNOWN CPTII MUTATIONS	1. **Dried blood spot card** (preferred) – 3 to 5 completely filled spots 2. **Whole Blood** – 1 mL of whole blood 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 per mutation**
CPT II DNA ANALYSIS – PANEL OF 8 MUTATIONS * The 8 Mutations included in panel are: • R124X • Q550R • Q413fs • P604S • P227L • R631C • L178F (534Tins/25bpdel) • S38fs (112-113 GC insertion) • DOES NOT INCLUDE THE COMMON MUTATION FOR CPTII (S113L) • MAY ORDER INDIVIDUAL MUTATIONS LISTED ON PANEL	4. **Whole Blood** (preferred) - 1 mL of whole blood 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 5. **Dried blood spot card** – 3 to 5 completely filled spots 6. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-6 weeks from receipt of sample	83890 83892 * 83894 * 83898 * 83912 *For each mutation requested*	Panel cost - $850.00 Individual mutation cost - $150.00

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CPTII ENZYME ASSAY*	1. **Fresh Whole Blood (preferred)** – 10 mL of peripheral blood collected aseptically in ACD tube (preferred) or Sodium Heparin tube. For the ACD tube, solution A or B is acceptable. A normal whole blood control must be submitted with the patient sample. The normal whole blood control should be collected, handled and shipped in the same manner as the patient sample. 2. **Fibroblasts** – 2 confluent T-25 flasks	Whole blood samples must be collected and shipped on the same day. In addition, the sample must be shipped overnight to preserve the integrity of the sample. **For all sample types, ship at room temperature Monday through Wednesday only, using a guaranteed overnight courier.**	Whole blood: 2-4 weeks from sample receipt Fibroblasts: 4-6 weeks from sample receipt	85999 (for blood) 88233 (for fibroblasts) 84157 82658	**$900.00**
DIABETES TYPE I INSULIN GENE (INS) MUTATION ANALYSIS BY DNA SEQUENCING	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-8 weeks from sample receipt	83890 83891 x 3 83892 83894 x 3 83898 x 3 83904 x 3 83909 x 3 83912	**$750.00**
DIABETES TYPE I INSULIN GENE (INS) DNA ANALYSIS FOR INDIVIDUAL KNOWN MUTATIONS	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2-4 weeks from sample receipt	83890 83891 83892 83894 83898 83904 83909 83912	**Individual mutation cost - $250.00**

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
FABRY DISEASE MUTATION ANALYSIS BY DNA SEQUENCING OF THE GLA GENE	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-8 weeks from sample receipt	83890 83891 x7 83892 83894 x7 83898 x7 83904 x7 83909 x7 83912	**$1,300.00**
FABRY DISEASE DNA ANALYSIS FOR INDIVIDUAL KNOWN MUTATIONS *	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2-4 weeks from sample receipt	83890 83891 83892 83894 83898 83904 83909 83912	**Individual mutation cost - $275.00**
LCHAD DNA ANALYSIS – COMMON MUTATION * (1528 G>C)	1. **Dried blood spot card (preferred)** 3 to 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**

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
FAMILIAL HYPERTROPHIC CARDIOMYOPATHY/ WOLFF-PARKINSON-WHITE SYNDROME – PRKAG2 GENE SEQUENCING	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Fibroblasts** - 2 confluent T-25 flasks 3. **Dried blood spot card** – 5 completely filled spots	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	3-4 weeks from receipt of sample	83890 83891 x16 83892 83894 x16 83898 x16 83904 x16 83909 x16 83912	$1,450.00
FAMILIAL HYPERTROPHIC CARDIOMYOPATHY/ WOLFF-PARKINSON-WHITE SYNDROME – INDIVIDUAL KNOWN MUTATION ANALYSIS	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 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 83891 83892 83894 83898 83904 83909 83912	$250.00
R302Q (905G>A) MUTATION ANALYSIS (WOLFF-PARKINSON-WHITE SYNDROME)	1. **Whole Blood (preferred)** - 6 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 – 3 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 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 83891 83892 83894 83898 83904 83909 83912	$250.00

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
MCAD DNA ANALYSIS– COMMON MUTATION * (985 A>G) OR OTHER INDIVIDUAL KNOWN MCAD MUTATIONS	1. **Dried blood spot card** <u>(preferred)</u> – 3 to 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 per mutation**
MCAD DNA ANALYSIS - PANEL OF 8 MUTATIONS * The 8 Mutations included in panel are: • 244insT • 583 G>A • 250C>T • 616 C>T • 617 G>A • 799 G>A • 503 A>G • 322-325 4bp del • DOES NOT INCLUDE THE COMMON MUTATION FOR MCAD (985A>G) • MAY ORDER INDIVIDUAL MUTATIONS LISTED ON PANEL	1. **Whole Blood** <u>(preferred)</u> - 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 2. **Dried blood spot card**– 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4 – 6 weeks from receipt of sample	83890 83892* 83894* 83898* 83912 *For each mutation requested*	**Panel cost - $850.00** **Individual mutation cost - $150.00**

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DNA SEQUENCING ANALYSIS FOR UNKNOWN CPTII OR MCAD MUTATIONS (IF DEFINITIVE DIAGNOSIS INFORMATION IS AVAILABLE, PLEASE ATTACH IT TO THE MOLECULAR CLINICAL TEST REQUISTION)	1. **Whole Blood (preferred)** - 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 is 0.5 mL 2. **Fibroblasts** - 2 confluent T-25 flasks 3. **Dried Blood spot card** - 5 completely filled spots	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	6-8 weeks from sample receipt	83890 83891 x12 83892 83894 x12 83898 x12 83904 x12 83909 x12 83912	$2,000.00
SALLA DNA ANALYSIS – COMMON MUTATION 115C>T (R39C) *	1. Whole Blood (preferred) - 3 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 – 1 mL 2. Dried blood spot card – 3 to 5 completely filled spots 3. Fibroblasts - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2 weeks from sample receipt	83890 83891 x2 83892 83894 x2 83898 x2 83904 x2 83909 x2 83912	$275.00
FREE SIALIC STORAGE DISORDERS (SASD) - UNKNOWN MUTATION ANALYSIS BY DNA SEQUENCING OF THE SLC17A5 GENE	1. **Whole Blood (preferred)** - 3 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 – 1 mL 2. **Dried blood spot card** –5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	4-8 weeks from sample receipt	83890 83891 x11 83892 83894 x11 83898 x11 83904 x11 83909 x11 83912	$2500.00

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TEST NAME	SAMPLE REQUIREMENTS	SHIPPING/HANDLING	TURNAROUND TIME	CPT CODE	COST
FREE SIALIC STORAGE DISORDERS (SASD) DNA ANALYSIS FOR INDIVIDUAL KNOWN MUTATIONS *	1. **Whole Blood (preferred)** - 3 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 – 1 mL 2. **Dried blood spot card** – 3 to 5 completely filled spots 3. **Fibroblasts** - 2 confluent T-25 flasks	For all sample types, ship overnight at room temperature using a guaranteed overnight courier.	2-4 weeks from sample receipt	83890 83891 83892 83894 83898 83904 83909 83912	**Individual mutation cost - $275.00**

* Prenatal diagnosis may be available as a research test for familial known mutations. Please contact the Molecular Diagnostics Laboratory Director or Quality Management for additional information.

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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	COST
AMINO ACIDS	1. **Plasma** (preferred) – 0.5 mL of heparinized plasma 2. **Serum** – 0.5mL of serum collected in a red-top tube 3. **CSF** – 0.5 mL collected in a sterile container and frozen at -80˚C **Absolute minimum for all sample types – 150 uL**	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	**$140.00**
HOMOCYSTEINE (total)	1. **Plasma**–0.5mL of heparinized plasma 2. **Serum** – 0.5 mL	**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	**$45.00**
LACTATE *Cerebrospinal Fluid*	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 receipt of sample.	83605	**$25.00**
5-METHYLTETRAHYDROFOLATE (5-MTHF) *Cerebrospinal Fluid*	0.5 ml – CSF; frozen at -80°C at collection. *See next sheet for CSF collection instructions*	**Call lab for SPECIAL Collection tubes** Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82491	**$125.00**
MONOAMINE NEUROTRANSMITTER METABOLITES (5-HIAA, HVA, 3-OMD) *Cerebrospinal Fluid*	0.5 ml – CSF; frozen at -80°C at collection. *See next sheet for CSF collection instructions*	**Call lab for SPECIAL Collection tubes** Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82492	**$125.00**
TETRAHYDROBIOPTERIN & NEOPTERIN PROFILE (BH4,N) *Cerebrospinal Fluid*	0.5 ml - CSF; frozen at -80°C at collection. *See next sheet for CSF collection instructions*	**Call lab for SPECIAL Collection tubes** Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82492	**$125.00**
NEOPTERIN *Cerebrospinal Fluid*	0.5 ml - CSF; frozen at -80°C at collection. *See next sheet for CSF collection instructions*	**Call lab for SPECIAL Collection tubes** Ship on 3 – 4 lbs. of dry ice using a guaranteed overnight courier	1 week from receipt of sample.	82491	**$65.00**

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

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	
1	0.5 mL	The total CSF volume required is 3.5 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, 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. 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.

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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	COST
MITOCHONDRIAL BETA OXIDATION DEFECTS	**Fibroblasts** – 2 sub-confluent T-25 flasks filled with medium supplemented with serum.	**Shipping:** 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. **Handling:** 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	88233 84157 83789	**$850.00**

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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 ☐ FEFEMALE

Medical Record # or Patient ID #: ____________________

Specimen Information – Sample 1

Accession/Lab ID # ____________

Specimen Type: ____________

Sample Date: ________ Time: ________

For Cell Lines- Flask Date: ________

Passage #: ________

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

Mass Spectrometry tests:

- ☐ Acylcarnitine Profile *
- ☐ Carnitine Levels *
- ☐ Organic Acids *
- ☐ Fabry Disease Screening *

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.

Specimen Information – Sample 2

Accession/Lab ID # ____________

Specimen Type: ____________

Sample Date: ________ Time: ________

For Cell Lines- Flask Date: ________

Passage #: ________

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

Mass Spectrometry tests:

- ☐ Acylcarnitine Profile *
- ☐ Carnitine Levels *
- ☐ Organic Acids *
- ☐ Fabry Disease Screening *

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.

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.

TO ASSIST INTERPRETATION PLEASE FILL IN BELOW

Primary Presenting Symptoms: ____________________

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Abnormal Labs: ____________________

Suspected Diagnosis: ____________________

Diet or Infant Formula: ____________________

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

RESULTS ADDRESS

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Phone: Fax:

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

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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*
- ☐ Homocysteine (total)*
- ☐ 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: ______________________________

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Abnormal Labs: ______________________________

Suspected Diagnosis: ______________________________

Diet or Infant Formula: ______________________________

Medications: ______________________________

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

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 .

RESULTS ADDRESS

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Phone: Fax:

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

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Phone: Fax:

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MOLECULAR DIAGNOSTICS CLINICAL TEST REQUISITION

PATIENT INFORMATION

Patient Name: ______________________________ Gender: ☐ Male ☐ Female

DOB or Age: ____________ Hospital or Patient ID: ____________________

Accession #: ____________________

SAMPLE INFORMATION

Sample type: ____________________ If fibroblasts, flask date: ________ passage#: ________

Sample date: ____________________ Sample Time: ____________________

CLINICAL HISTORY:

Symptoms or Clinical findings: ____

Family history: ____

Ordering Physician:	Phone:	Fax:

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TEST(S) REQUESTED:

☐ Alpha-1 Antitrypsin Deficiency – Common Mutation (E342K) Analysis

☐ Alpha-1 Antitrypsin Deficiency – Panel of 8 mutations or individual mutation (check ALL for panel)

- ☐ Glu342Lys ☐ Pro386 Ser
- ☐ Val361Ala ☐ Glu400Asp
- ☐ Leu362Met ☐ Gln401Lys
- ☐ Gly373Trp ☐ Thr416Pro

☐ Alpha-1 Antitrypsin Deficiency – SERIPINA1 Mutation Analysis by DNA Sequencing

☐ Alpha-1 Antitrypsin Deficiency – SERIPINA1 Individual known mutation analysis. Indicate mutation: ____________

☐ CPTII DNA analysis – Common Mutation (S113L)

☐ CPTII DNA analysis – Panel of 8 mutations or individual mutation (check ALL for panel)

- ☐ R124X ☐ Q550R
- ☐ Q413fs ☐ P604S
- ☐ P227L ☐ R631C
- ☐ L178F (534Tins/25bpdel) ☐ S38fs (112-113 GC insertion)

☐ CPTII DNA Analysis for individual known mutation. Indicate mutation: ____________

☐ CPTII Enzyme Assay

☐ CPTII – Unknown Mutation Analysis by DNA sequencing

☐ Diabetes Type I Insulin Gene (INS) Mutation Analysis by DNA Sequencing

☐ Diabetes Type I Insulin Gene (INS) Individual known mutation analysis. Indicate mutation: ____________

RESULTS ADDRESS

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Phone: Fax:

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

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Phone: Fax:

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MOLECULAR DIAGNOSTICS CLINICAL TEST REQUISITION

PATIENT INFORMATION

Patient Name: ______________________ Gender: ☐ Male ☐ Female

DOB or Age: ____________ Hospital or Patient ID: ______________

Accession #: ______________

SAMPLE INFORMATION

Sample type: ______________ If fibroblasts, flask date: _______ passage#: _______

Sample date: ______________ Sample Time: ______________

CLINICAL HISTORY:

Symptoms or Clinical findings: ______________________

Family history: ______________________

Ordering Physician:	Phone:	Fax:

<u>NY State Clientele MUST check AT LEAST one of the following</u>: [ ] 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:

☐ EIF2B -Related Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter Disease (CACH/VWM)

- O Complete DNA Sequencing of the EIF2B Gene
- O Sequencing (INDICATE SUBUNIT BELOW):
  - ☐ EIF-2B1 - alpha
  - ☐ EIF-2B2 – beta
  - ☐ EIF-2B3 - gamma
  - ☐ EIF-2B4 - delta
  - ☐ EIF-2B5 – epsilon
- O Common mutation R113H + panel of 50 known mutations
- O Target or Carrier mutation analysis. Indicate mutation: ______________

☐ Fabry Disease - Unknown Mutation Analysis by DNA sequencing of the GLA gene

☐ Fabry Disease DNA Analysis for individual known mutation. Indicate mutation: ______________

☐ Familial Hypertrophic Cardiomyopathy/Wolff-Parkinson-White Syndrome - PRKAG2 Gene Sequencing

☐ Familial Hypertrophic Cardiomyopathy/Wolff-Parkinson-White Syndrome - R302Q (905G>A) Mutation Analysis

☐ Familial Hypertrophic Cardiomyopathy/Wolff-Parkinson-White Syndrome – individual known mutation analysis

Indicate mutation: ______________

<u>RESULTS ADDRESS</u>

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Phone: Fax:

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Phone: Fax:

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MOLECULAR DIAGNOSTICS CLINICAL TEST REQUISITION

PATIENT INFORMATION

Patient Name: ______________________ Gender: ☐ Male ☐ Female

DOB or Age: ____________ Hospital or Patient ID: ______________________

Accession #: ______________________

SAMPLE INFORMATION

Sample type: ______________________ If fibroblasts, flask date: ________ passage#: ________

Sample date: ______________________ Sample Time: ______________________

CLINICAL HISTORY:

Symptoms or Clinical findings: ______________________

Family history: ______________________

Ordering Physician:	Phone:	Fax:

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TEST(S) REQUESTED:

☐ LCHAD DNA Analysis– Common Mutation (1528G>C)

☐ MCAD DNA analysis - Common Mutation (985A>G)

☐ MCAD DNA analysis – Panel of 8 mutations or individual mutation (check ALL for panel)

☐ 244insT	☐ 583 G>A
☐ 250C>T	☐ 616 C>T
☐ 322-325 4bp del	☐ 617 G>A
☐ 503 A>G	☐ 799 G>A

☐ MCAD DNA Analysis for individual known mutation. Indicate mutation: ______________________

☐ Unknown MCAD Mutation Analysis by DNA sequencing

☐ Salla DNA analysis – Common mutation 115 C>T (R39C)

☐ SASD Disease DNA Analysis for individual known mutation. Indicate mutation: ______________________

☐ Unknown SASD Mutation analysis by DNA sequencing of SLC17A5 gene

RESULTS ADDRESS

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Phone: Fax:

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Phone: Fax:

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WEBSITE: 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 CPTII Enzyme Assay and 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.