

**ROBERT GUTHRIE BIOCHEMICAL & MOLECULAR GENETICS LABORATORY**

**Georgirene D. Vladutiu, Ph.D., Director**

THE BUFFALO GENERAL HOSPITAL

KALEIDA HEALTH

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www.rgbgl.org

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Biochemical Analysis  
for Metabolic Myopathy  
Page 1 of 4

**REQUEST FOR LABORATORY ANALYSIS**

**PATIENT INFORMATION**

Please include Your accession # NP \_\_\_\_\_

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone: \_\_\_\_\_

Sex \_\_\_\_\_ DOB \_\_\_\_\_ Age \_\_\_\_\_ Race \_\_\_\_\_

Ethnicity \_\_\_\_\_

Diagnostic Code(s) - **Required** \_\_\_\_\_

**REFERRING PHYSICIAN INFORMATION:**

Date of Request: \_\_\_\_\_

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

E-Mail: \_\_\_\_\_

Mail Results to: Referring Physician   
Or Institution   
Or other physician

**INSTITUTION INFORMATION:** (For billing and resulting):

Guarantor: \_\_\_\_\_

Contact Person: \_\_\_\_\_

Department: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone: \_\_\_\_\_

**OTHER PHYSICIAN INFORMATION:**

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone: \_\_\_\_\_

Fax: \_\_\_\_\_

The referring institution will be billed.

**SPECIMEN INFORMATION:**

Tissue Submitted:  
[ ] Whole Blood  
[ ] Skeletal Muscle (type) \_\_\_\_\_  
[ ] Cardiac Muscle  
[ ] Liver  
[ ] Other: \_\_\_\_\_

Tissue Source:  
[ ] Autopsy  
[ ] Biopsy

Specimen Collection Date: \_\_\_\_\_

Specimen Storage Prior to Shipment:  
[ ] Liquid Nitrogen  
[ ] -70° C  
[ ] -20° C

This patient has consented to the testing of his/her tissue or body fluid for an inborn error of metabolism.  
The implications of genetic testing have been discussed with the patient. **(Required)**

Physician's initials \_\_\_\_\_  
Date: \_\_\_\_\_

<b>OFFICE USE ONLY</b>	
Date Received	Amount Received
ID Number	Comments
Initials	

Patient Name: \_\_\_\_\_ Current Medications & Dosage: \_\_\_\_\_  
Reason for Referral: \_\_\_\_\_ Current Supplements: \_\_\_\_\_  
\_\_\_\_\_ Cholesterol-Lowering Drugs \_\_\_\_\_  
\_\_\_\_\_ And Dosage (if applicable): \_\_\_\_\_

Please ✓ appropriate choices

FAMILY HISTORY		GENERAL CHARACTERISTICS		NEUROMUSCULAR ABNORMALITIES				
<input type="checkbox"/>	50	Consanguinity in family	<input type="checkbox"/>	1760	Respiratory distress	<input type="checkbox"/>	6051	Headache
<input type="checkbox"/>	51	On cholesterol-lowering drug	<input type="checkbox"/>	1761	Multiple congenital anomalies	<input type="checkbox"/>	6052	Migraine
<input type="checkbox"/>	100	Mental retardation	<input type="checkbox"/>	1765	Hypoglycemia	<input type="checkbox"/>	6075	Visual loss
<input type="checkbox"/>	125	Psychomotor retardation	<input type="checkbox"/>	1770	Cerebral palsy	<input type="checkbox"/>	7000	Ophthalmoplegia
<input type="checkbox"/>	150	Metabolic disorder, hx of	<input type="checkbox"/>	1775	Visual impairment	<input type="checkbox"/>	7050	Rhabdomyolysis
<input type="checkbox"/>	175	Viral infection, assn. with	<input type="checkbox"/>	1777	Behavior problems	<input type="checkbox"/>	8000	Heat stroke
<input type="checkbox"/>	200	Affected child	<input type="checkbox"/>	1801	Neurofibromatosis	<input type="checkbox"/>	8050	Malignant hyperthermia
<input type="checkbox"/>	201	Affected parent	<input type="checkbox"/>	1802	Fascio-scapulo-humeral disease	<input type="checkbox"/>	8075	<b>Normal</b> neurologic exam
<input type="checkbox"/>	202	Affected sibling	<input type="checkbox"/>	1805	Diabetes	<input type="checkbox"/>	8076	Abnormal neurologic exam
<input type="checkbox"/>	250	Ethnic predilection	<b>NEUROMUSCULAR ABNORMALITIES</b>					
<input type="checkbox"/>	275	<b>No</b> family hx of present illness	<input type="checkbox"/>	2000	Muscle pain	<b>LABORATORY STUDIES</b>		
<input type="checkbox"/>	280	Hearing impairment	<input type="checkbox"/>	2050	Muscle weakness	<input type="checkbox"/>	10000	Metabolic acidosis
<input type="checkbox"/>	281	Diabetes, history of	<input type="checkbox"/>	3000	Muscle cramps	<input type="checkbox"/>	10025	<b>No</b> metabolic acidosis
<b>GENERAL CHARACTERISTICS</b>		<input type="checkbox"/>	3010	Fibromyalgia	<input type="checkbox"/>	10030	Respiratory acidosis	
<input type="checkbox"/>	1000	Hepatomegaly	<input type="checkbox"/>	3025	Fatigue	<input type="checkbox"/>	10050	Hypoglycemia
<input type="checkbox"/>	1025	Hepatitis	<input type="checkbox"/>	3030	Lethargy	<input type="checkbox"/>	10055	<b>No</b> hypoglycemia
<input type="checkbox"/>	1026	Liver failure	<input type="checkbox"/>	3050	Stiffness	<input type="checkbox"/>	10075	Neutropenia
<input type="checkbox"/>	1027	Hepatic dysfunction	<input type="checkbox"/>	3060	Abnormal gait	<input type="checkbox"/>	10076	Hemolytic anemia
<input type="checkbox"/>	1050	Splenomegaly	<input type="checkbox"/>	3065	History of falling	<input type="checkbox"/>	10100	Hyperammonemia
<input type="checkbox"/>	1100	Cardiomegaly	<input type="checkbox"/>	3066	Clumsiness	<input type="checkbox"/>	10125	<b>No</b> hyperammonemia
<input type="checkbox"/>	1125	Cardiomyopathy	<input type="checkbox"/>	3075	Ataxia	<input type="checkbox"/>	10150	Lactic acidemia
<input type="checkbox"/>	1140	Other cardiac abnormalities	<input type="checkbox"/>	3080	Dystonia	<input type="checkbox"/>	10155	<b>Normal</b> lactate
<input type="checkbox"/>	1145	Kidney abnormalities	<input type="checkbox"/>	3085	Spastic diplegia	<input type="checkbox"/>	10160	Lactate unknown
<input type="checkbox"/>	1146	Renal failure	<input type="checkbox"/>	3090	Dysphagia	<input type="checkbox"/>	10200	Ketosis
<input type="checkbox"/>	1147	Gastrointestinal problems	<input type="checkbox"/>	3092	Dysarthria	<input type="checkbox"/>	10225	<b>No</b> ketosis
<input type="checkbox"/>	1148	Hyperthyroidism	<input type="checkbox"/>	4000	Exercise intolerance	<input type="checkbox"/>	10250	Elevated CK: Unit:
<input type="checkbox"/>	1149	Hypothyroidism	<input type="checkbox"/>	4001	<b>Normal</b> EEG	<input type="checkbox"/>	10260	<b>Normal</b> CK
<input type="checkbox"/>	1150	Unusual hair or nails	<input type="checkbox"/>	4004	Abnormal EEG	<input type="checkbox"/>	10265	CK unknown
<input type="checkbox"/>	1175	Apnea	<input type="checkbox"/>	4025	<b>Normal</b> brain MRI	<input type="checkbox"/>	10266	Elevated aldolase
<input type="checkbox"/>	1200	Developmental delay	<input type="checkbox"/>	4005	Abnormal brain MRI/CT	<input type="checkbox"/>	10267	<b>Normal</b> aldolase
<input type="checkbox"/>	1215	Microcephaly	<input type="checkbox"/>	4007	<b>Normal</b> EMG	<input type="checkbox"/>	10275	Elevated CSF protein
<input type="checkbox"/>	1221	Motor delay	<input type="checkbox"/>	4010	Abnormal EMG	<input type="checkbox"/>	10276	Elevated CSF lactate
<input type="checkbox"/>	1222	Motor regression	<input type="checkbox"/>	4012	<b>Normal</b> EKG	<input type="checkbox"/>	10277	<b>Normal</b> CSF protein
<input type="checkbox"/>	1225	Short stature	<input type="checkbox"/>	4013	Abnormal EKG	<input type="checkbox"/>	10280	Elevated liver enzymes
<input type="checkbox"/>	1250	Coarse facies	<input type="checkbox"/>	4015	Demyelination	<input type="checkbox"/>	10285	<b>Normal</b> liver enzymes
<input type="checkbox"/>	1275	Dysmorphic features	<input type="checkbox"/>	4017	Nerve dysfunction	<input type="checkbox"/>	10286	Iron deposition in hepatocytes
<input type="checkbox"/>	1276	Congenital malformations	<input type="checkbox"/>	4019	<b>Normal</b> ischemic exercise test	<input type="checkbox"/>	10300	Elevated pyruvate
<input type="checkbox"/>	1300	Corneal clouding	<input type="checkbox"/>	4020	Abnormal ischemic exercise test	<input type="checkbox"/>	10310	<b>Normal</b> pyruvate
<input type="checkbox"/>	1325	Retinal degeneration	<input type="checkbox"/>	4030	Encephalopathy	<input type="checkbox"/>	10315	Pyruvate unknown
<input type="checkbox"/>	1350	Cataracts	<input type="checkbox"/>	4040	Microcephaly	<input type="checkbox"/>	10316	Lactate/pyruvate >25
<input type="checkbox"/>	1375	Skeletal anomalies	<input type="checkbox"/>	4050	Myoglobinuria	<input type="checkbox"/>	10318	Lactate/pyruvate <25
<input type="checkbox"/>	1400	Umbilical hernia	<input type="checkbox"/>	4051	<b>No</b> pigmenturia	<input type="checkbox"/>	10320	Lactate/pyruvate normal
<input type="checkbox"/>	1425	Inguinal hernia	<input type="checkbox"/>	4052	Myoglobinemia	<input type="checkbox"/>	10325	Elevated plasma acylcarnitine
<input type="checkbox"/>	1450	Failure to thrive	<input type="checkbox"/>	5000	Seizures	<input type="checkbox"/>	10330	Plasma carnitine deficiency
<input type="checkbox"/>	1500	Vomiting	<input type="checkbox"/>	5001	Tremor	<input type="checkbox"/>	10335	<b>Normal</b> plasma carnitine
<input type="checkbox"/>	1550	Recurrent Infections	<input type="checkbox"/>	5050	Stroke	<input type="checkbox"/>	10350	Elevated urine organic acids
<input type="checkbox"/>	1600	Fasting-induced symptoms	<input type="checkbox"/>	6000	Myoclonus	<input type="checkbox"/>	10355	<b>Normal</b> urine organic acids
<input type="checkbox"/>	1625	<b>Normal</b> intelligence	<input type="checkbox"/>	6025	Choreoathetoid movements	<input type="checkbox"/>	10360	Abnormal plasma amino acids
<input type="checkbox"/>	1650	Intellectual impairment	<input type="checkbox"/>	6026	Hypertonia	<input type="checkbox"/>	10366	Abnormal urinary mucopolysaccharides
<input type="checkbox"/>	1660	<b>Normal</b> hearing	<input type="checkbox"/>	6027	Parkinsonism			
<input type="checkbox"/>	1675	Hearing impairment	<input type="checkbox"/>	6028	Decreased tone	<input type="checkbox"/>	10365	<b>Normal</b> plasma amino acids
<input type="checkbox"/>	1700	Progressive course	<input type="checkbox"/>	6030	<b>Normal</b> ophthalmologic exam	<input type="checkbox"/>	10370	Chromosome abnormality Type:
<input type="checkbox"/>	1725	Non-progressive course	<input type="checkbox"/>	6035	<b>Abnormal</b> ophthalmologic exam			
<input type="checkbox"/>	1750	Hypotonia	<input type="checkbox"/>	6050	Ptosis	<input type="checkbox"/>	10371	<b>Normal</b> chromosomes

Patient Name: \_\_\_\_\_

Please ✓ appropriate choices

MOLECULAR STUDIES			MUSCLE HISTOLOGY	
<input type="checkbox"/>	10375	MELAS mutation present	<input type="checkbox"/>	11281 Ring Fibers
<input type="checkbox"/>	10376	MELAS mutation absent	<input type="checkbox"/>	11285 Vacuolar myopathy
<input type="checkbox"/>	10377	MERRF mutation present	<input type="checkbox"/>	11286 Rimmed vacuoles
<input type="checkbox"/>	10378	MERRF mutation absent	<input type="checkbox"/>	11287 Nemaline rods
<input type="checkbox"/>	10379	mtDNA deletion present	<input type="checkbox"/>	11280 <b>Normal</b> biopsy
<input type="checkbox"/>	10380	mtDNA deletion absent	<b>MUSCULAR HISTOCHEMISTRY</b>	
<input type="checkbox"/>	10381	NARP mutation present	<input type="checkbox"/>	12000 Phosphorylase absent
<input type="checkbox"/>	10382	NARP mutation absent	<input type="checkbox"/>	12025 Phosphorylase present
<input type="checkbox"/>	10383	LHON mutation present	<input type="checkbox"/>	12050 Myoadenylate deaminase absent
<input type="checkbox"/>	10384	LHON mutation absent	<input type="checkbox"/>	12055 Myoadenylate deaminase present
<input type="checkbox"/>	13230	CPT2 mutation present	<input type="checkbox"/>	12100 Cytochrome c oxidase absent
<input type="checkbox"/>	13231	CPT2 mutation absent	<input type="checkbox"/>	12125 Cytochrome c oxidase present
<input type="checkbox"/>	13240	Myophosphorylase mutation present	<input type="checkbox"/>	12400 Cytochrome c oxidase increased
<input type="checkbox"/>	13241	Myophosphorylase mutation absent	<input type="checkbox"/>	12150 Succinate dehydrogenase absent
<input type="checkbox"/>	13250	Myoadenylate deaminase mutation present	<input type="checkbox"/>	12175 Succinate dehydrogenase present
<input type="checkbox"/>	13151	Myoadenylate deaminase mutation absent	<input type="checkbox"/>	12410 Succinate dehydrogenase increased
<input type="checkbox"/>	13152	Other mutation: Specify	<input type="checkbox"/>	12200 Phosphofructokinase absent
<b>MUSCLE HISTOLOGY</b>			<input type="checkbox"/>	12225 Phosphofructokinase present
<input type="checkbox"/>	11000	Ragged red fibers	<input type="checkbox"/>	12275 Lipid storage absent
<input type="checkbox"/>	11010	<b>No</b> ragged red fibers	<input type="checkbox"/>	12250 Lipid storage present
<input type="checkbox"/>	11012	Increased trichrome staining	<input type="checkbox"/>	12325 Glycogen storage absent
<input type="checkbox"/>	11025	Esterase-positive fibers	<input type="checkbox"/>	12300 Glycogen storage present
<input type="checkbox"/>	11050	Fiber type grouping	<input type="checkbox"/>	12350 NADH reactions absent
<input type="checkbox"/>	11070	Type 1 fiber predominance	<input type="checkbox"/>	12375 NADH reactions present
<input type="checkbox"/>	11075	Type 2 fiber predominance	<input type="checkbox"/>	12450 NADH reactions increased
<input type="checkbox"/>	11100	Fiber atrophy	<input type="checkbox"/>	12376 NADH: targetoid
<input type="checkbox"/>	11105	Fiber hypertrophy	<input type="checkbox"/>	12377 NADH: moth eaten
<input type="checkbox"/>	11125	Fiber size variation	<b>ELECTRON MICROSCOPY</b>	
<input type="checkbox"/>	11150	Central cores	<input type="checkbox"/>	13000 Membrane-bound glycogen
<input type="checkbox"/>	11175	Myopathic features	<input type="checkbox"/>	13050 Glycogen pools
<input type="checkbox"/>	11180	Neuropathic features	<input type="checkbox"/>	13051 <b>Normal</b> glycogen
<input type="checkbox"/>	11185	Inflammatory infiltrates	<input type="checkbox"/>	13100 Abnormal number mitochondria
<input type="checkbox"/>	11200	Fiber necrosis/regeneration	<input type="checkbox"/>	13125 Abnormal structure mitochondria
<input type="checkbox"/>	11250	Denervating process	<input type="checkbox"/>	13130 <b>Normal</b> mitochondria
<input type="checkbox"/>	11265	Few internal nuclei	<input type="checkbox"/>	13150 Inclusions absent
<input type="checkbox"/>	11270	Moderate internal nuclei	<input type="checkbox"/>	13155 Inclusions present
<input type="checkbox"/>	11275	Many internal nuclei	<input type="checkbox"/>	13165 Vacuoles absent
<input type="checkbox"/>	11276	<b>No</b> increase in internal nuclei	<input type="checkbox"/>	13160 Vacuoles present
			<input type="checkbox"/>	13166 Lipid droplets
			<input type="checkbox"/>	13176 Acute necrosis
			<input type="checkbox"/>	13177 <b>No</b> abnormalities

I, the referring physician, will provide for genetic counseling of this patient if a hereditary disorder is identified as a result of the requested testing to insure that the results and implications are understood by the patient. (Required)

Referring Physician's Signature: \_\_\_\_\_ Date: \_\_\_\_\_

Patient Name: \_\_\_\_\_

Please ✓ appropriate choices

**MITOCHONDRIAL MYOPATHY PROFILE**  
**Tissue Requirement: 125 mgs  
or 25 mgs/test (skeletal or cardiac muscle)**

- NADH dehydrogenase
- NADH cytochrome c reductase
- Succinate dehydrogenase
- Succinate cytochrome c reductase
- Cytochrome c oxidase
- Citrate synthase

**FATTY ACID TRANSPORT**  
**Tissue Requirement: 50 mgs/test**

- Carnitine, free and esterified
- Carnitine palmitoyltransferase (CPT) + citrate synthase

**MODIFIED MITOCHONDRIAL MYOPATHY PROFILE**  
**Tissue Requirement: 25 mgs/test**

- Succinate dehydrogenase
- Cytochrome c oxidase
- Citrate synthase

**COENZYME Q10**  
**Tissue Requirement: 100 mgs**  
**Signed consent form required. (See "Forms")**

**FUMARASE**  
**Tissue Requirement: 25 mgs**

**GLYCOGEN STORAGE DISEASE PROFILE**  
**Tissue Requirement: 200 mgs (skeletal or cardiac muscle)**

- Acid and neutral maltase
- Phosphorylase
- Phosphorylase b kinase
- Phosphofructokinase

**MYOGLOBINURIA PROFILE**  
**Tissue Requirement: 250 mgs or 25 mgs/test  
unless otherwise noted**

- Phosphorylase
- Phosphorylase b kinase
- Phosphofructokinase
- Phosphoglycerate kinase
- Phosphoglycerate mutase
- Carnitine palmitoyltransferase (CPT) (50 mgs)  
+ citrate synthase
- Lactate dehydrogenase
- Myoadenylate deaminase (50 mgs)

**Shipping and Handling:** Tissue should be snap frozen in liquid nitrogen at the time of biopsy, stored at -70°C until shipment, then sent to the laboratory on dry ice by overnight carrier. Please notify the laboratory by telephone at the time of shipment.