

ROBERT GUTHRIE BIOCHEMICAL & MOLECULAR GENETICS LABORATORY

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THE BUFFALO GENERAL HOSPITAL

KALEIDA HEALTH

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CLIA License #33D0685375 • New York State Laboratory Permit # PF12005

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: _____

OFFICE USE ONLY

| | | | |
|---------------|-------|-----------------|-------|
| Date Received | _____ | Amount Received | _____ |
| ID Number | _____ | Comments | _____ |
| Initials | _____ | | _____ |

Patient Name: _____
Reason for Referral: _____

Current Medications & Dosage: _____
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 | Normal 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 | NEUROMUSCULAR ABNORMALITIES | | | LABORATORY STUDIES | | |
| <input type="checkbox"/> | 275 | No family hx of present illness | <input type="checkbox"/> | 2000 | Muscle pain | <input type="checkbox"/> | 10000 | Metabolic acidosis |
| <input type="checkbox"/> | 280 | Hearing impairment | <input type="checkbox"/> | 2050 | Muscle weakness | <input type="checkbox"/> | 10025 | No metabolic acidosis |
| <input type="checkbox"/> | 281 | Diabetes, history of | <input type="checkbox"/> | 3000 | Muscle cramps | <input type="checkbox"/> | 10030 | Respiratory acidosis |
| GENERAL CHARACTERISTICS | | | <input type="checkbox"/> | 3010 | Fibromyalgia | <input type="checkbox"/> | 10050 | Hypoglycemia |
| <input type="checkbox"/> | 1000 | Hepatomegaly | <input type="checkbox"/> | 3025 | Fatigue | <input type="checkbox"/> | 10055 | No hypoglycemia |
| <input type="checkbox"/> | 1025 | Hepatitis | <input type="checkbox"/> | 3030 | Lethargy | <input type="checkbox"/> | 10075 | Neutropenia |
| <input type="checkbox"/> | 1026 | Liver failure | <input type="checkbox"/> | 3050 | Stiffness | <input type="checkbox"/> | 10076 | Hemolytic anemia |
| <input type="checkbox"/> | 1027 | Hepatic dysfunction | <input type="checkbox"/> | 3060 | Abnormal gait | <input type="checkbox"/> | 10100 | Hyperammonemia |
| <input type="checkbox"/> | 1050 | Splenomegaly | <input type="checkbox"/> | 3065 | History of falling | <input type="checkbox"/> | 10125 | No hyperammonemia |
| <input type="checkbox"/> | 1100 | Cardiomegaly | <input type="checkbox"/> | 3066 | Clumsiness | <input type="checkbox"/> | 10150 | Lactic acidemia |
| <input type="checkbox"/> | 1125 | Cardiomyopathy | <input type="checkbox"/> | 3075 | Ataxia | <input type="checkbox"/> | 10155 | Normal lactate |
| <input type="checkbox"/> | 1140 | Other cardiac abnormalities | <input type="checkbox"/> | 3080 | Dystonia | <input type="checkbox"/> | 10160 | Lactate unknown |
| <input type="checkbox"/> | 1145 | Kidney abnormalities | <input type="checkbox"/> | 3085 | Spastic diplegia | <input type="checkbox"/> | 10200 | Ketosis |
| <input type="checkbox"/> | 1146 | Renal failure | <input type="checkbox"/> | 3090 | Dysphagia | <input type="checkbox"/> | 10225 | No ketosis |
| <input type="checkbox"/> | 1147 | Gastrointestinal problems | <input type="checkbox"/> | 3092 | Dysarthria | <input type="checkbox"/> | 10250 | Elevated CK Unit: |
| <input type="checkbox"/> | 1148 | Hyperthyroidism | <input type="checkbox"/> | 4000 | Exercise intolerance | <input type="checkbox"/> | 10260 | Normal CK |
| <input type="checkbox"/> | 1149 | Hypothyroidism | <input type="checkbox"/> | 4001 | Normal EEG | <input type="checkbox"/> | 10265 | CK unknown |
| <input type="checkbox"/> | 1150 | Unusual hair or nails | <input type="checkbox"/> | 4004 | Abnormal EEG | <input type="checkbox"/> | 10266 | Elevated aldolase |
| <input type="checkbox"/> | 1175 | Apnea | <input type="checkbox"/> | 4005 | Abnormal brain MRI/CT | <input type="checkbox"/> | 10267 | Normal aldolase |
| <input type="checkbox"/> | 1200 | Developmental delay | <input type="checkbox"/> | 4007 | Normal EMG | <input type="checkbox"/> | 10275 | Elevated CSF protein |
| <input type="checkbox"/> | 1215 | Microcephaly | <input type="checkbox"/> | 4010 | Abnormal EMG | <input type="checkbox"/> | 10276 | Elevated CSF lactate |
| <input type="checkbox"/> | 1221 | Motor delay | <input type="checkbox"/> | 4012 | Normal EKG | <input type="checkbox"/> | 10277 | Normal CSF protein |
| <input type="checkbox"/> | 1222 | Motor regression | <input type="checkbox"/> | 4013 | Abnormal EKG | <input type="checkbox"/> | 10280 | Elevated liver enzymes |
| <input type="checkbox"/> | 1225 | Short stature | <input type="checkbox"/> | 4015 | Demyelination | <input type="checkbox"/> | 10285 | Normal liver enzymes |
| <input type="checkbox"/> | 1250 | Coarse facies | <input type="checkbox"/> | 4017 | Nerve dysfunction | <input type="checkbox"/> | 10286 | Iron deposition in hepatocytes |
| <input type="checkbox"/> | 1275 | Dysmorphic features | <input type="checkbox"/> | 4019 | Normal ischemic exercise test | <input type="checkbox"/> | 10300 | Elevated pyruvate |
| <input type="checkbox"/> | 1276 | Congenital malformations | <input type="checkbox"/> | 4020 | Abnormal ischemic exercise test | <input type="checkbox"/> | 10310 | Normal pyruvate |
| <input type="checkbox"/> | 1300 | Corneal clouding | <input type="checkbox"/> | 4025 | Normal brain MRI | <input type="checkbox"/> | 10315 | Pyruvate unknown |
| <input type="checkbox"/> | 1325 | Retinal degeneraton | <input type="checkbox"/> | 4030 | Encephalopathy | <input type="checkbox"/> | 10316 | Lactate/pyruvate >25 |
| <input type="checkbox"/> | 1350 | Cataracts | <input type="checkbox"/> | 4050 | Myoglobinuria | <input type="checkbox"/> | 10318 | Lactate/pyruvate <25 |
| <input type="checkbox"/> | 1375 | Skeletal anomalies | <input type="checkbox"/> | 4051 | No pigmenturia | <input type="checkbox"/> | 10320 | Lactate/pyruvate normal |
| <input type="checkbox"/> | 1400 | Umbilical hernia | <input type="checkbox"/> | 4052 | Myoglobinemia | <input type="checkbox"/> | 10325 | Elevated plasma acylcarnitine |
| <input type="checkbox"/> | 1425 | Inguinal hernia | <input type="checkbox"/> | 5000 | Seizures | <input type="checkbox"/> | 10330 | Plasma carnitine deficiency |
| <input type="checkbox"/> | 1450 | Failure to thrive | <input type="checkbox"/> | 5001 | Tremor | <input type="checkbox"/> | 10335 | Normal plasma carnitine |
| <input type="checkbox"/> | 1500 | Vomiting | <input type="checkbox"/> | 5050 | Stroke | <input type="checkbox"/> | 10350 | Elevated urine organic acids |
| <input type="checkbox"/> | 1551 | Recurrent Infections | <input type="checkbox"/> | 6000 | Myoclonus | <input type="checkbox"/> | 10355 | Normal urine organic acids |
| <input type="checkbox"/> | 1600 | Fasting-induced symptoms | <input type="checkbox"/> | 6025 | Choreoathetoid movements | <input type="checkbox"/> | 10360 | Abnormal plasma amino acids |
| <input type="checkbox"/> | 1625 | Normal intelligence | <input type="checkbox"/> | 6026 | Hypertonia | <input type="checkbox"/> | 10366 | Abnormal urinary mucopolysaccharides |
| <input type="checkbox"/> | 1650 | Intellectual impairment | <input type="checkbox"/> | 6027 | Parkinsonism | <input type="checkbox"/> | 10365 | Normal plasma amino acids |
| <input type="checkbox"/> | 1660 | Normal hearing | <input type="checkbox"/> | 6028 | Decreased tone | <input type="checkbox"/> | 10370 | Chromosome abnormality |
| <input type="checkbox"/> | 1675 | Hearing impairment | <input type="checkbox"/> | 6030 | Normal ophthalmologic exam | | | Type: |
| <input type="checkbox"/> | 1700 | Progressive course | <input type="checkbox"/> | 6035 | Abnormal ophthalmologic exam | <input type="checkbox"/> | 10371 | Normal chromosomes |
| <input type="checkbox"/> | 1725 | Non-progressive course | <input type="checkbox"/> | 6050 | Ptosis | | | |
| <input type="checkbox"/> | 1750 | Hypotonia | | | | | | |

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 | Normal biopsy |
| <input type="checkbox"/> | 10380 | mtDNA deletion absent | MUSCULAR HISTOCHEMISTRY | | |
| <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 |
| MUSCLE HISTOLOGY | | | <input type="checkbox"/> | 12225 | Phosphofructokinase present |
| <input type="checkbox"/> | 11000 | Ragged red fibers | <input type="checkbox"/> | 12275 | Lipid storage absent |
| <input type="checkbox"/> | 11010 | No 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 | ELECTRON MICROSCOPY | | |
| <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 | Normal 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 | Normal 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 | No 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 | No 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: 200 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

PURINE METABOLISM DEFECTS
Tissue Requirement: 50 mgs
● Myoadenylate deaminase

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.