



5424 Glenridge Drive NE
Atlanta, GA 30342
678.225.0222 FAX 678.225.0212
www.mnglab.com

SPECIMEN COLLECTION & PROCESSING REQUIREMENTS

John M Shoffner MD, Medical Director CLIA License # 11D0703390 State of Georgia License # 044-146

**Please ship all specimens overnight in insulated containers at the proper temperature for receipt by our lab Monday – Friday.
(No Saturday/Sunday deliveries accepted)**

For assistance, please call 678-225-0220 or see our website at www.mnglab.com

METABOLIC TESTING (CSF, BLOOD, URINE)

FROZEN SPECIMENS: Ship on freezer packs (-20°C) or dry ice (-70°C) for next day 10am delivery.

Plasma Amino Acids: Collect 1-4 ml of blood in sodium heparin (green top) tube. Although not required, sample collection after a 3-4 hour fast is optimal. Draw off plasma. Store frozen at -20°C and ship frozen. Minimum volume of plasma: 0.5 ml.

Serum or Plasma Carnitine: Collect 2-4 ml of blood in red top tube (serum) or green top tube (sodium heparin) for plasma. Although not required, sample collection after a four hour fast is optimal. Draw off serum or plasma. Store frozen at -20°C and ship frozen. Minimum volume: 1 ml serum or plasma.

Blood Lactate: Collect 2-4 ml blood in sodium fluoride (gray top) tube (3 ml). Remove plasma and freeze at -20°C. Minimum plasma volume: 0.5 ml. Store frozen at -20°C and ship frozen.

CoQ10 Plasma: Collect 2-4 ml blood in sodium heparin (green top) or EDTA (purple top) tube. Remove plasma and freeze at -20°C. Minimum plasma volume: 0.5 ml. Store frozen at -20°C and ship frozen.

CSF Amino Acids: Collect 1 ml of fluid. Spin sample (remove red blood cells) and freeze the clear CSF. Specimens may be maintained frozen for no more than 7 days prior to processing. Minimum volume: 1.0 ml CSF. Store frozen at -20°C and ship frozen.

CSF Lactate: Collect 1 ml of fluid. Spin sample (remove red blood cells) and freeze the clear CSF. Specimens may be maintained frozen for no more than 7 days prior to processing. Minimum volume: 0.5 ml CSF. Store frozen at -20°C and ship frozen.

CSF 5-Methyltetrahydrofolate: Collect 1 ml of fluid. Spin sample (remove red blood cells) and freeze the clear CSF at -70°C. Minimum volume: 0.5 ml CSF. **Store frozen at -70°C and ship on 5-10 lbs dry ice for next day 10am delivery.**

CSF Succinyladenosine: Collect 1 ml of fluid. Spin sample (remove red blood cells) and freeze the clear CSF at -70°C. Minimum volume: 0.5 ml CSF. **Store frozen at -70°C and ship on 5-10 lbs dry ice for next day 10am delivery.**

CSF Sialic Acid: Collect 1 ml of fluid. Spin sample (remove red blood cells) and freeze the clear CSF at -70°C. Minimum volume: 0.5 ml CSF. **Store frozen at -70°C and ship on 5-10 lbs dry ice for next day 10am delivery.**

Thymidine and Deoxyuridine (plasma): Collect 2-4 ml blood in sodium heparin (green top) or EDTA (purple top) tube. Remove plasma and freeze at -20°C. Minimum plasma volume: 0.5 ml. Store frozen at -20°C and ship frozen.

Urine organic acids and amino acids: Collect 5-10 ml of a random sample. Do not add preservatives. Specimens may be maintained frozen for no more than 14 days prior to processing. Minimum urine volume: 3 ml. Store frozen at -20°C and ship frozen.

Aromatic L-Amino Acid Decarboxylase Enzymology (plasma): Collect 2-4 ml of blood in EDTA (purple top) or sodium heparin (green top) tube. Remove plasma and freeze at -70°C; Minimum plasma volume: 0.5 ml. **Store frozen at -70°C and ship on 5-10 lbs dry ice for next day 10am delivery.**

PYRUVATE processing instructions (Blood and CSF):

Blood Pyruvate: Collect 2-4 ml of blood in a green top (Sodium Heparin) (Green Top) Immediately after collection, add 2 ml COLD 8% perchloric acid per 1 ml of specimen. Vortex 30 seconds, place on ice for 5 minutes. Spin down, draw off and freeze supernatant. Store frozen at -20°C and ship frozen. Minimum volume: 1 ml whole blood

CSF Pyruvate: Collect 1 ml of fluid. Immediately after collection, add 2 ml COLD 8% perchloric acid per 1 ml of specimen. Vortex 30 seconds, place on ice for 5 minutes. Minimum volume: 1.0 ml CSF. Store frozen at -20°C and ship frozen.

ROOM TEMPERATURE BLOOD SPECIMENS: Ship in insulated container for next day delivery.

CoQ10 (Leukocyte): Collect 5 ml of whole blood in acid-citrate-dextrose (ACD) tube (Yellow top). Maximum stability is 5 days. Minimum volume 2-5 ml blood. Ship samples collected within last 24 hrs in insulated container at room temperature. Blood collected on Friday is discouraged as maximum stability is 5 days. If Friday or holiday collection occurs, hold at 4°C and ship on ice within 3 days; do NOT freeze. EDTA (purple top) tube may be used but is discouraged as stability is limited to 48 hours.

Thymidine Phosphorylase Enzymology (leukocyte): Collect 8-10 ml whole blood in acid-citrate-dextrose (ACD) tube (Yellow top). Maximum stability is 5 days. Minimum volume 2-5 ml blood. Ship samples collected within last 24 hrs in insulated container at room temperature. Blood collected on Friday is discouraged as maximum stability is 5 days. If Friday or holiday collection occurs, hold at 4°C and ship on ice within 3 days; do NOT freeze. EDTA (purple top) tube may be used but is discouraged as stability is limited to 48 hours.

MUSCLE HISTOLOGY TESTING (MUSCLE)

Histochemistry and Immunohistochemistry: Tissue size: ≥ 0.5 gm (1 CM³) Requires specialized collection protocols. Please call our laboratory with questions or visit website at www.mnglab.com. Note that this sample size is appropriate for histochemistry/immunohistochemistry reactions only. (Oxidative phosphorylation enzymology requires a larger sample size.)

To Send Fresh Sample: Place specimen in a securely closed container (such as Petri dish) that contains sterile gauze moistened with normal saline. It is very important NOT to submerge the specimen in saline or to have excessively wet gauze. PLACE THE SPECIMEN CONTAINER ON ICE.

Specimen MUST arrive at Medical Neurogenetics laboratory within 2 hours.

To Send Frozen Sample: Samples must be frozen at the time of biopsy in ISOPENTANE cooled by liquid nitrogen. Keep sample frozen at -80°C. Ship overnight, 5-10 lbs dry ice.

To Send Muscle in Formalin: It is critical that specimen is kept at room temperature until placed into formalin. Send formalin specimen cold (ROOM TEMP) same day or overnight.



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OXPHOS ENZYMOLOGY (MUSCLE)

Tissue size: ≥ 1 gm (1.5-2.0 CM³) No electrocautery near sample. This is approximately the size of the tip of the little finger (5th finger) measured from the base of the nail bed.

Fresh Muscle: (Biopsy must be scheduled through Medical Neurogenetics office) Place specimen in a securely closed container (such as Petri dish) that contains sterile gauze moistened with normal saline. It is very important not to submerge the specimen in saline or to have excessively wet gauze. PLACE THE SPECIMEN CONTAINER ON ICE but do NOT freeze.

Must arrive at Medical Neurogenetics within 2 hours of collection.

Frozen Muscle: Place tissue immediately in liquid nitrogen (or isopentane cooled with liquid nitrogen). Keep sample frozen at -80°C .

Ship overnight, 5-10 lbs dry ice.

PROTEIN CHEMISTRY TESTING (MUSCLE)

MUSCLE TISSUE for Western Blot Testing: SHIP OVERNIGHT, 5-10 LBS DRY ICE.

Tissue Size: Minimum ≥ 5 mm³ per individual protein chemistry test ordered. Place tissue immediately in liquid nitrogen (or isopentane cooled with liquid nitrogen).

Keep sample frozen at -80°C .

Ship overnight, 5-10 lbs dry ice.

MOLECULAR/DNA TESTING (MUSCLE, BLOOD, EXTRACTED DNA, URINE, BUCCAL SWAB KIT)

FROZEN SPECIMENS: Ship on 5-10 lbs dry ice for next day 10am delivery.

MUSCLE or other TISSUES for DNA Testing:

Tissue Size: Minimum ≥ 5 mm³. Place tissue immediately in liquid nitrogen (or isopentane cooled with liquid nitrogen). Keep sample frozen at -80°C .

Ship overnight, 5-10 lbs dry ice.

ROOM TEMPERATURE SPECIMENS: Ship in insulated container for next day delivery.

Blood Samples for DNA Testing: Collect blood in EDTA (purple top) tube.

Minimum Volumes: Adult: 8-10 ml; Children: 5-10 ml; Infants: 2-5 ml.

Ship samples collected within last 24 hrs in insulated container at room temperature.

Blood collected on Friday is discouraged as maximum stability is 5 days. If Friday or holiday collection occurs, hold at 4°C and ship on ice within 3 days; do NOT freeze.

Urine Epithelial Cells for mtDNA Sequencing: Collect 20 ml of a random sample. Do not add preservatives.

Minimum Volume: 20mls urine. Sample should ship within 24 hours of collection.

Ship samples collected within last 24 hrs in insulated container at room temperature.

Buccal Swab for mtDNA sequencing: Sample Collection Buccal Swab Kit is supplied with instructions by MNG. Please call 678-225-0222 ext 109 for kits to be shipped to your facility.

RESPIROMETRY

Fresh Muscle: Tissue size: ≥ 0.3 gm (≥ 5 mm³). Biopsy MUST BE scheduled through the Medical Neurogenetics office (678.225.0222 ext 103) and performed in Atlanta.

Fibroblast Specimens: Send two T-25 flasks filled with 13% FBS/DMEM media (90% confluency). Cell lines must be certified free from Mycoplasma – send confirmatory test results with flasks.

Ship flasks at room temperature filled with media in well-insulated container at room temperature.

Before shipping, call Tissue Culture Lab at 678-225-0222 Ext. 110 to schedule an appointment for testing.

TISSUE CULTURE TESTING

For specimens below, call Medical Neurogenetics 678-225-0222 ext 110 prior to shipping. Specimens must arrive within 24 hours of collection. No sample receipt on Fridays or weekends.

Tissue Culture of skin biopsy: Skin biopsy may be taken from the inner forearm with a sterile skin punch (2-5mm) or at incision site of muscle biopsy. **Site is sterilized with alcohol swabs only** (other reagents will inhibit cell isolation/growth). Biopsy site may be anaesthetized with 2% lidocaine with epinephrine. Depth of punch is 2mm. Transfer skin biopsy into 15ml conical filled with sterile culture medium (13%FBS/DMEM with 1% antibiotic). Cell culture media may be requested from MNG.

Ship specimen on ice (**do not freeze**) overnight.

Any deviations from this processing must be approved by Medical Neurogenetics staff.

EBV Lymphocyte Transformation: 10 cc EDTA whole blood.

Send overnight, room temperature in insulated container.

Fibroblast Specimens: Send two T-25 flasks filled with culture medium (13% FBS/DMEM media) at 90% confluency. MUST BE certified free from mycoplasma.

Ship flasks at room temperature in well-insulated container.

NEUROCHEMISTRY TESTING

CSF requires specialized collection protocols & tubes. See separate Neurochemistry Collection and Processing Protocol on our website at www.mnglab.com. For CSF collection kits, please contact the Neurochemistry Department at 678-225-0222 ext 125 or by emailing your request to support@mnglab.com.