

Protocol for Metabolic Disorder Autopsy

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Reviewed and approved by _____ Pathology Department
Steiner RD, Cederbaum, Stephen D. Laboratory evaluation of urea cycle disorders. J Pediatr 2001; 138(1): S28-29.

* To be performed within 4 to 6 hrs of death if at all possible *

**1. Call the Newborn Screening Laboratory of the Oregon Health Division
(503) 229-5466**

If the infant was born in the state of Washington: #(206) 361-2902

- Ask the lab to hold any newborn screening blood spots that they might still be holding and not to discard them (Oregon's lab currently holds newborn screening cards for up to one year)

2. Obtain a skin biopsy from the Achilles tendon: place in RPMI (keep at room temperature)

- Take to Central Lab (3rd floor D. H. Hall) ASAP where fibroblasts can be grown in culture
- Any questions should be directed to cytogenetics on call

3. Collect blood from cardiac (atrium) puncture: place in the following tubes (~5 ml ea.) :

* To Central Lab, ASAP *

- **1 red top** (small) – have lab spin and store serum at -70°
- **3 sodium heparin tubes** (usually green tops)
 - 1 green top, lab spin and store at -70°
 - 1 green top, 4°C (on wet ice), to biochemical genetics lab for separation and isolation of white blood cells
 - 1 green top, at room temperature to cytogenetics lab
- **1 EDTA** (lavender top) – leave unspun, at 4°C (on wet ice), to biochemical genetics lab
- **1 ACD tube** (yellow)- FOR DNA – keep at 4°C and send it to the DNA diagnostics lab

These tubes can be used to order the tests below and others, depending on clinical situation:

Blood amino acids

Blood lactate-may not be accurate post-mortem

Blood carnitine level and acylcarnitine profile

Blood lysosomal storage disease screening

Blood sterols

Comprehensive metabolic panel, cholesterol, triglycerides, uric acid, ammonia – may not be accurate postmortem

Urine organic acids

Urine metabolic screen

Urine MPS and oligosaccharides

4. Fill four filter paper spots on a newborn screening card

5. **Collect bile by direct puncture of the gallbladder and store frozen (2-3 ml is sufficient)**
 - If VL. is smaller, the bile can be spotted on filter paper and stored at room temp.
6. **Urine** can be collected by suprapubic tap or traces of urine may be collected from the posterior bladder wall with a cotton swab. If unable to collect urine, collect **vitreous humor** from the eye with a syringe & freeze in liquid nitrogen (keep at -70°) [vitreous is good for organic acid analysis]
7. **Collect several grams of muscle:** cut into several 2 gram pieces- place in labeled aluminum foil, and freeze in liquid nitrogen or dry ice (keep at -70°)
 - This is for mitochondrial disease testing *** **Call 4-5400**, BCGENLAB for questions
8. **Collect several 2-gr pieces of liver, brain, kidney, and heart muscle** with the same technique and place into separate containers.
 - Also, collect each of these tissues for **routine histology** and **electron microscopy** (glutaraldehyde)

METABOLIC DISEASE CONTACT:

Robert Steiner, MD (email: Steinerr@ohsu.edu), Phone: 4-7859
Or, call 4-9000 and ask for the Metabolic Disease Specialist on call

Reference Text (for further information on Metabolic Disease):

The Metabolic and Molecular Bases of Inherited Disease, 8th ed., 2000.