

DEPARTMENT OF PATHOLOGY AND LABORATORY MEDICINE
BIOCHEMICAL GENETICS

Instructions for Collecting, Handling & Shipping Specimens

Address for Shipping, Mailing and Couriers:

Please use ONLY FedEx or UPS - DO NOT SHIP VIA POSTAL SERVICE

Children's Hospital Colorado
Clinical Lab - Biochemical Genetics
13123 East 16th Ave, B120 - Room B0200
Aurora CO 80045

All specimens must be received with an appropriate test requisition form that has been fully completed to include the following information:

- Name of patient
- Date of birth of patient
- Collection date and time for specimen(s)
- Name of physician ordering the test
- Name, address and contact phone number of facility requesting the test
- Your facility accession number for each specimen (for your reference)
- Test(s) requested - one form may be used for requesting multiple tests

Quantitative Tests (no patient preparation required)

All samples should be frozen immediately (and kept frozen). Samples should be sent overnight on dry ice in a well-sealed Styrofoam container. Mail samples early in the week to avoid weekend arrival.

- **Plasma or serum acylcarnitine profile:** 0.2 mL heparinized plasma collected in green-topped tubes (or serum), frozen and sent on dry ice.
- **Serum or plasma amino acids:** 0.3 mL serum (or heparinized plasma collected in green-topped tubes), frozen and sent on dry ice.
- **Cerebrospinal fluid (CSF) amino acids:** Minimum volume 0.1 mL (at least 0.3 mL preferred), frozen and sent on dry ice.
- **Urine amino acids:** 2.0 mL random urine, frozen and sent on dry ice.
- **Urine and/or serum glutaric acid, 3-hydroxyglutaric acid, methylmalonic acid, orotic acid, hexanoylglycine, phenylpropionylglycine and succinylacetone (stable isotope dilution):** 0.5 mL serum (or heparinized plasma) and/or 2.0 mL urine, frozen.
- **Tay-Sachs disease carrier status:** 0.3 mL serum (or heparinized plasma), sent with a serum from a control (non-Jewish male with no Tay-Sachs disease in family) if possible.

Qualitative Screening Tests (no patient preparation required)

All samples should be sent in a well-constructed tube with a secure lid to prevent leakage during transport. Freeze urine or CSF immediately without preservative.

- **Serum or plasma amino acids:** 0.3 mL serum (or heparinized plasma collected in green-topped tubes), ambient temperature.
- **Urine amino or organic acids:** 2.0 mL random urine, no preservative, ambient temperature.
- **Urine mucopolysaccharides:** 10.0 mL random urine, no preservative, ambient temperature.
- **Urine or CSF Bratton-Marshall Test (succinylpurines):** 2.0 mL CSF or random morning urine. Freeze urine or CSF immediately without preservative.

Urine Trimethylaminuria (TMAu) Testing: Trimethylamine (TMA) and Trimethylamine-n-oxide (TMAO)

- TMAu Collection Information and Shipping and Handling Instructions

Leukocyte Enzyme Assay (with or without mutation analysis) for VLCADD

- VLCADD Collection Information and Shipping and Handling Instructions

Fibroblast GA1 and Pyruvate Dehydrogenase Deficiency (PCDC) Assays

- Please send two T-25 flasks containing confluent fibroblasts (with plug-seal caps, completely filled with media) in a Styrofoam box at room temperature.
- Fibroblasts must be mycoplasma free and grown only in fetal calf serum of U.S. origin.
- A test requisition form must be filled out for each patient and sent with the specimens. Please also include the passage number (if known) and any relevant information about the patient.