

Biochemical Genetics Laboratory 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 – Rm. 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)

- Plasma or serum acylcarnitine profile:
0.2 mL heparinized plasma collected in green-topped tubes (or serum), frozen and sent on dry ice. **MUST INCLUDE PATIENT DOB.**
- Serum or plasma amino acids:
0.3 mL serum (or heparinized plasma collected in green-topped tubes), frozen and sent on dry ice. Include patient age.
- Cerebrospinal fluid (CSF) amino acids:
Minimum volume 0.1 mL (at least 0.3 mL preferred), frozen and sent on dry ice. Include patient age.
- Urine amino acids:
2.0 mL random urine, frozen and sent on dry ice. Include patient age.
- Urine and/or serum glutaric acid, 3-hydroxyglutaric acid, methylmalonic acid, orotic acid, hexanoylglycine, phenylpropionylglycine, N-acetylaspartic acid 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.

Serum should be frozen immediately, packed in dry ice in a well-sealed styrofoam container and sent overnight delivery. Mail samples early in the week to avoid weekend arrival.

All samples should be kept frozen until sent. Samples should be sent on dry ice in a well-constructed tube with a secure lid to prevent leakage during transport. Samples should not be sent in urine collection cups.

Qualitative Screening tests (no patient preparation required)

- Serum or plasma amino acids:
0.3 mL serum (or heparinized plasma collected in green-topped tubes), ambient temperature.
- Urine amino acids:
2.0 mL random urine, no preservative, ambient temperature.
- Urine 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 urine (morning urine specimen preferred).

Freeze urine or CSF immediately without preservative.

All samples should be sent in a well-constructed tube with a secure lid to prevent leakage during transport. Samples should not be sent in urine collection cups.

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

- For collection instructions please see the TMAu Testing Inquiries section on our website.

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

Please choose the appropriate printer-friendly (PDF) document:

- [VLCADD collection information \(PDF file\)](#)

Fibroblast GA1, GA2 (MADD), 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.

Primary contact for fibroblast specimens, tissue culture and GA1 & GA2 (MADD) testing:

Dr. Michael Woontner

Phone: 720-777-0506

Email: Michael.Woontner@ChildrensColorado.org