

NOTE: For additional information and resources, please call the Rare Diseases Information Center at **1-888-205-2311**, or visit their website at: <http://rarediseases.info.nih.gov/GARD/>.

TRIMETHYLAMINE IN URINE BY GAS CHROMATOGRAPHY

PRINCIPLE:

Urine, to which an internal standard is added, is made alkaline and heated in a sealed tube, releasing volatile basic and neutral compounds into the head-space of the tube. The gaseous phase is sampled via gas-tight syringe and injected into a gas chromatographic system with a flame-ionization detector and amine separating column. Quantitation is effected by similar treatment of aqueous trimethylamine standards, comparing peak area ratios to concentration.

Trimethylaminuria is presumably a genetic defect in the enzymatic conversion of trimethylamine (odor of stale fish) to trimethylamine-N-oxide (non-odiferous). It causes body odor severe enough to create serious psychosocial problems. Diagnosis is made by detection of excess trimethylamine in urine. Available therapies include dietary restriction of choline (precursor of TMA) and also dietary restriction of foods containing inhibitors of TMA → TMA-oxide enzyme (brassicac, tea, spices).

SPECIMENS:

A. Patient preparation:

1. None for random specimens; send random specimen if patient is symptomatic (choline load will increase symptoms).
2. Instructions for choline load (patient non-symptomatic):
 - a. Collect a random specimen and label "pre-load" (see specimen handling procedure below).
 - b. Give 50 mg choline/Kg body weight, not to exceed 3g choline bitartrate. (Choline bitartrate can be found in health food stores).
 - c. Collect entire specimen between 6-12 hours following choline dose; label "post-load".
 - d. Keep refrigerated during specimen collection.
 - e. Mix well and send a 15-20 mL aliquot for testing (see below).

B. Specimen:

1. **15-20 mL** random urine and/or aliquot of post-choline load specimen.
2. *Adjust urine to **pH ~5.0** using a small volume of concentrated HCl.
3. *Fill several **glass** tubes (glass screw-cap tubes or glass red-top vacutainer tubes) no more than **half full** to allow for expansion of the specimen due to freezing.
4. *Freeze, cushion tubes well (gauze or bubble-wrap) to minimize breakage, and send by overnight express with **5-6 lbs of dry ice** in a styrofoam box. Please ship Monday - Wednesday only.
5. **Trimethylamine is a highly volatile compound and will be lost from the specimen during shipment if these instructions are not followed! Urine samples must be kept tightly sealed and frozen in glass tubes between collection and analysis of TMA; there is evidence that air shipment of samples in plastic allows loss of TMA.**

REFERENCE: Brewster, MA, Schedewie, H. Trimethylaminuria. ANNALS OF CLIN. AND LAB. SCIENCE 1983; 13(1): 20-24.

SHIPPING INSTRUCTIONS:

1. Ship on dry ice by Federal Express or DHL Express, priority overnight, Mon. – Wed. only, to:

Clinical Laboratory
Arkansas Children's Hospital
1 Children's Way
Little Rock, AR 72202
Attention: Lab Receiving
Charge for assay: \$140.65
Turnaround time: 6-8 weeks

FAX: (501) 364-4222
Phone: (501) 364-1300
CPT Codes: 82491, 80500

2. **PLEASE COMPLETE INFORMATION FORM AND SEND WITH PATIENT'S SPECIMEN.**

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