



## Biochemical Genetics Laboratory

The Biochemical Genetics Laboratory in the Department of Pediatrics at the University of California San Diego has been in operation since the establishment of the UCSD Medical School in 1969. We offer tests not generally available which have been developed in the conduct of research or the care of our patients, and endeavor to assure the maximum quality and reliability. The goal of the UCSD Biochemical Genetics Laboratory is to provide comprehensive diagnostic laboratory services to assist in the diagnosis and treatment of inborn errors of metabolism. A special feature of our lab is our consultation services with health care professionals who specialize in inborn errors of metabolism, and a D.V.M, Ph.D. is available to you for consultation on our laboratory results. Please feel free to call if we can be of assistance in your diagnostic or therapeutic plans.

### Shipping Address:

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### General Information:

- Laboratory working hours are Monday through Friday, 08:00 to 16:00 PST. As our regular hours do not include weekends or holidays, we request that specimens be shipped routinely Monday-Thursday.
- Use only guaranteed overnight carriers (U.S. Postal Express may take longer and specimens may be ruined).
- Please label each specimen with patient's name and date/time of collection, using permanent ink, and place in a resealable plastic biohazard bag, one per bag (gummed labels fall off frozen specimens). Please place completed test request form in separate plastic bag to avoid contamination from specimen.
- **For International clients, payment arrangements must be received before processing specimen(s).**

**Laboratory Director:** G. Diane Shelton, D.V.M., Ph.D. California License #: 6987

Certification numbers: CLIA ID # O5D0643075 Federal Tax ID # 33-0833316  
California Dept. of Health Services Clinical Laboratory License, ID # CLE4102  
College of American Pathologists (CAP) #2318702

Proficiency Test Programs: College of American Pathologists  
Veterinarian Affiliations: American College of Veterinary Internal Medicine

Following is a list of tests offered for veterinary patients by our laboratory.

<b>Test</b>	<b>Specimens</b>
AMINO ACIDS, QUANTITATIVE ANALYSIS	Plasma, Urine
ORGANIC ACIDS, QUANTITATIVE COMPREHENSIVE	Urine
METHYLMALONIC ACID (MMA) QUANTITATION	Urine
ACYLCARNITINE PROFILE	Plasma
TAURINE, QUANTITATIVE ANALYSIS	Plasma
HOMOCYST(E)INE, TOTAL	Plasma
CARNITINE, QUANTITATIVE ANALYSIS	Urine, Plasma, Tissue
METABOLIC PANEL	Plasma, Urine

The following pages summarize the individual tests and specify the sample requirements, turn-around times and prices.

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**QUANTITATIVE AMINO ACID ANALYSIS****(Urine, Plasma)**

Comments: Standard analysis using modern automated amino acid analyzer. We will resolve any ambiguities using an independent, two column analyzer. Please be aware that for accurate determination of homocystine in blood, special handling is required - you may call our lab to arrange specifically for plasma homocystine.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice), or lyophilized and shipped at room temp with original volume specified.  
Plasma, ≥1 mL (minimum 0.5 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice) or lyophilized and shipped at room temperature with original volume specified.

Turn-around time: Routine: 14-21 working days

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**ORGANIC ACIDS QUANTITATIVE COMPREHENSIVE****(Urine)**

Comments: This analysis is useful in the evaluation of dogs with persistent lactic acidemia and/or lipid storage myopathy. Evaluations are also essential for dogs with unexplainable neurological disorders, failure to thrive, or elevated anion gaps. Our organic acid analysis is based on a state-of-the-art application of gas chromatography-mass spectrometry. Identification of metabolites is definitive, and we fully quantitate more than 150 compounds. Note that other laboratories may give qualitative results only or report results without mass spectral identification, which can lead to serious misinterpretation.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice) or lyophilized and shipped at room temp with original volume specified.

Turn-around time: Routine: 14-21 working days

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**QUANTITATIVE METHYLMALONIC ACID (MMA) ANALYSIS****(Urine)**

Comments: Quantitation of MMA excretion can be used to follow the clinical status and therapeutic response of patients with methylmalonic acidemia or vitamin B<sub>12</sub> deficiency. We use gas chromatography-mass spectrometry for definitive identification and precise quantitation.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice) or lyophilized and shipped at room temp with original volume specified.

Turn-around time: Routine: 14-21 working days

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**QUANTITATIVE ACYLCARNITINE ANALYSIS****(Plasma)**

Comments: Acylcarnitines are assayed by tandem mass spectrometry. Reported values are accompanied with interpretation of profile. Acylcarnitines are very useful in diagnosing mitochondrial fatty acid beta oxidation disorders and several other organic acidemias.

Sample requirements: Plasma, ≥1 mL (0.5 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice or lyophilized).

Turn-around time: Routine: 14-21 working days.

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**QUANTITATIVE TAURINE ANALYSIS****(Plasma)**

Comments: Taurine is important in animal nutrition and it is vital in the developing newborn. We assay taurine using an automated amino acid analyzer.

Sample requirements: Plasma,  $\geq 1$  mL (minimum 0.5 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice) or lyophilized and shipped at room temperature with original volume specified.

Turn-around time: Routine: 14-21 weeks.

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**HOMOCYST(E)INE, TOTAL****(Plasma)**

Comments: Elevation of homocysteine is diagnostic for homocystinuria, and has been associated with vaso-occlusive and thrombotic disease. Where older methods only measured free homocystine, this method by tandem massspectrometry detects total (free and protein-bound, reduced homocysteine and oxidized homocystine). Please note that this new method does detect homocyst(e)ine in healthy subjects, so the normal range is not directly comparable to that of older methods.

Sample requirements: Plasma, 1 mL (minimum 0.5 mL) (separated from heparinized or EDTA-treated blood (green or purple-top tube), promptly frozen and shipped frozen (packed with dry ice) by overnight carrier.

Turn-around time: 14-21 working days.

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**QUANTITATIVE CARNITINE ANALYSIS****(Urine, Plasma, Tissue)**

Comments: Carnitine is assayed using carnitine acyl transferase, with and without alkaline hydrolysis of esters. Carnitine deficiency can be seen in various myopathies, and secondarily in a number of mitochondrial and metabolic diseases. Reported values are total, free, and esterified carnitine.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice) or lyophilized and shipped at room temp with original volume specified.

Plasma,  $\geq 1$  mL (0.5 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice) or lyophilized and shipped at room temperature with original volume specified.

Tissue, at least 1 gram muscle, rapidly frozen at  $-70^{\circ}\text{C}$ , stored and shipped frozen (packed with dry ice).

Turn-around time: Routine: 14-21 working days.

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**METABOLIC PANEL (PLASMA QUANTITATIVE AMINO ACIDS AND URINE QUANTITATIVE ORGANIC ACID SCREEN)****(Urine and Plasma)**

Comments: We offer these two tests at a special panel price. Please refer above for test details.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice) or lyophilized and shipped at room temp with original volume specified.

Plasma,  $\geq 1$  mL (minimum 0.5 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice) or lyophilized and shipped at room temperature with original volume specified.

Turn-around time: Routine: 14-21 working days.

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**Prices**

<b>Service</b>	<b>Price (US \$)</b>
<b>AMINO ACIDS, QUANTITATIVE ANALYSIS</b>	<b>165.</b>
<b>ORGANIC ACIDS, QUANTITATIVE COMPREHENSIVE</b>	<b>200.</b>
<b>METHYLMALONIC ACID (MMA) QUANTITATION</b>	<b>72.</b>
<b>ACYLCARNITINE PROFILE</b>	<b>197.</b>
<b>TAURINE</b>	<b>78.</b>
<b>HOMOCYST(E)INE, TOTAL</b>	<b>66.</b>
<b>CARNITINE, QUANTITATIVE ANALYSIS: PLASMA, URINE</b>	<b>78.</b>
<b>CARNITINE, QUANTITATIVE ANALYSIS: TISSUE</b>	<b>131.</b>
<b>METABOLIC PANEL</b>	<b>365.</b>