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 these M.D., Ph.D.'s are available to you for interpretation. 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.
- In the event of medical emergency, special shipping arrangements can be made, and we are generally able to perform testing in the fastest possible time, including weekends and holidays.
- 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.

Certification numbers:	CLIA ID # O5D0643075	Federal Tax ID # 95-6006144
	MediCal Lab # Lab 04102F	MediCare # 55L0008759
	California Dept. of Health Services Clinical Laboratory License, ID # CLE4102	
	College of American Pathologists (CAP) #2318702	
Proficiency Test Programs:	American Association of Bioanalysts, College of American Pathologists	
Physician Affiliations:	American Board of Medical Genetics, American Board of Pediatrics	

Following is a list of tests offered by our laboratory.

Test	Specimens
ACYLCARNITINE PROFILE	**Plasma**
AMINO ACIDS, QUANTITATIVE ANALYSIS	**Plasma, Urine, CSF**
CARBOXYLASES (PYRUVATE, PROPIONYL-COA, AND 3-METHYLCROTONYL-COA)	**Blood, Cultured Cells**
CARNITINE, QUANTITATIVE ANALYSIS	**Urine, Plasma, Tissue**
FIBROBLAST/AMNIOCYTE CULTURE SERVICES	**Biopsy, Cultured Cells**
HOMOCYST(E)INE, TOTAL	**Plasma**
HYPOXANTHINE-GUANINE/ADENINE PHOSPHORIBOSYL TRANSFERASE	**Bloodspots**
METHYLMALONIC ACID (MMA) QUANTITATION	**Urine, Plasma**
***N*-ACETYLASPARTATE QUANTITATION**	**Urine**
ORGANIC ACIDS, QUANTITATIVE COMPREHENSIVE	**Urine, Plasma, CSF**
PKU PANEL (PHENYLALANINE AND TYROSINE)	**Bloodspots**
OROTIC ACID QUANTITATION	**Urine**
SUCCINYLACETONE QUANTITATION	**Urine**
SUCCINYLPURINE SCREEN	**Urine**

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

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

Comments: Standard analysis using modern automated amino acid 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), .
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) .
Cerebrospinal fluid, ≥1 mL (minimum 0.5 mL)[standard plastic LP tube or transferred to red top tube], frozen and shipped frozen (packed with dry ice).

Turn-around time: Routine: 3-5 working days, Stat: same day.

QUANTITATIVE ORGANIC ACID COMPREHENSIVE **(Urine, Plasma, Cerebrospinal Fluid)**

Comments: 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).
Plasma, ≥2 mL (minimum 1.0 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice).
Cerebrospinal fluid, ≥2 mL (minimum 1.0 mL)[standard plastic LP tube or transferred to red top tube], frozen and shipped frozen (packed with dry ice).

Turn-around time: Routine: 5-7 working days, Stat: 2 days.

QUANTITATIVE OROTIC ACID ANALYSIS **(Urine)**

Comments: Orotic aciduria may be seen in deficiency of orotate decarboxylase, or following a single dose of allopurinol in females who are carriers for ornithine transcarbamylase deficiency. We use gas chromatography-mass spectrometry for definitive identification.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice).

Turn-around time: Routine: 5-7 working days, Stat: 2 days.

QUANTITATIVE *N*-ACETYLASPARTATE ANALYSIS **(Urine)**

Comments: Gross elevations of N-acetyl aspartate can be used to diagnose Canavan disease. We use gas chromatography-mass spectrometry for definitive identification.

Sample requirements: Urine, 10-20 mL (minimum 5 mL), frozen without preservatives and shipped frozen (packed with dry ice).

Turn-around time: Routine: 5-7 working days, Stat: 2 days.

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

Comments: Quantitation of MMA excretion can be used to follow the clinical status and therapeutic response of patients with methylmalonic acidemia. 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).
Plasma, ≥2 mL (minimum 1.0 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice).

Turn-around time: Routine: 5-7 working days, Stat: 2 days.

QUANTITATIVE CARNITINE ANALYSIS, FREE AND TOTAL **(Urine, Plasma, Tissue)**

Comments: Carnitine is assayed by tandem mass spectrometry, with and without alkaline hydrolysis of esters. 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).
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).
Tissue, at least 30 mg muscle, rapidly frozen at -70°C, stored and shipped frozen (packed with dry ice).

Turn-around time: Routine: 10-14 working days. Please provide information about carnitine therapy or a delay of an additional 10-14 working days could result for repeat analysis.

QUANTITATIVE ACYLCARNITINE ANALYSIS **(Plasma)**

Comments: Acylcarnitines are assayed by tandem mass spectrometry. Reported values are 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: 5-7 working days.
Please provide information about carnitine therapy and clinical history to assist with interpretation.

SET-UP OF FIBROBLAST CULTURE **(Biopsy)**

Comments: We can establish a culture of your patient's fibroblasts in order to assay in our lab or to convey to a reference lab for a particular assay or genetic test.

Sample requirements: Biopsy (skin or other specimen with adequate connective tissue), usually a single 3 or 4 mm diameter piece of skin extending to the epidermal-dermal junction is sufficient, kept in sterile medium at room temperature and shipped overnight.

Turn-around time: Variable, depending on sample origin and condition, generally 4-6 week minimum.

FIBROBLASTCONTINUED CULTURE **(Cultured Cells)**

Comments: When we receive flasks of cultured cells, we continue to grow them until analysis is completed and then they are discarded, unless other arrangements are made.

Sample requirements: Cultured cells (fibroblasts), two T-25 flasks on hand in our lab or shipped overnight with medium at room temperature.

Turn-around time: Depends on analysis and on sample condition.

FIBROBLAST STORAGE **(Cultured Cells)**

Comments: Long-term storage of cultured cells in liquid nitrogen (9-12 ampules). Storage guaranteed for one year, unless other arrangements are made.

Sample requirements: Cultured cells previously grown in our lab.

Turn-around time: Not applicable.

FIBROBLAST RECULTURED FROM STORAGE **(Cultured Cells)**

Comments: Cultured cells which are stored can be thawed and recultured for further analysis.

Sample requirements: Cultured cells (previously grown and stored by our laboratory).

Turn-around time: Variable, depending on sample origin and condition, generally 4-6 week minimum.

HYPOXANTHINE-GUANINE/ADENINE PHOSPHORIBOSYL TRANSFERASE **(Blood Spots)**

Comments: The diagnosis of Lesch-Nyhan syndrome and variant forms depends on the assay of HPRT, and the diagnosis of 2,8-dihydroxyadenine nephrolithiasis depends on assay of APRT. Our assay is radiochemical, and we measure both transferases as internal controls. It is helpful to have a clinical history of the patient provided.

Sample requirements: Blood Spots, PKU card (Guthrie Card Filter Paper) fill at least 3 spots, allow to air dry for 4 hours. Mail in envelope at room temperature.

Turn-around time: 10-14 working days.

SUCCINYLPURINE SCREEN **(Urine)**

Comments: Succinylpurine products (succinylAICAriboside [sAICAr] and succinyladenosine [sAdo]) are elevated in the body fluids of patients with deficiency of adenylosuccinate lyase, a disorder which can present with mental retardation, seizures and autistic features. We use tandem mass spectrometry to detect sAICAr and sAdo and report qualitative results, identifying samples with elevated amounts or with abnormal ratios. This method is not subject to the false positives which complicate Bratton-Marshall screening.

Sample requirements: Urine, 10-20 mL (min 2) frozen and shipped on dry ice.

Turn-around time: 10-14 working days.

SUCCINYLACETONE QUANTITATION **(Urine, Plasma)**

Comments: Succinylacetone excretion can be used to follow the clinical status and therapeutic response of patients with tyrosinemia (type 1, hepatorenal). 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) .

Turn-around time: Routine: 5-7 working days, Stat: 2 days.

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: 10-14 working days.

PKU PANEL (PHENYLALANINE AND TYROSINE) **(Bloodspots)**

Comments: Standard analysis using modern automated amino acid analyzer or tandem mass spectrometry. Used to monitor dietary management of phenylketonuria.

Sample requirements: Blood Spots, PKU card (Guthrie Card Filter Paper) fill at least 3 spots, allow to air dry for >4 hours. Mail in envelope at room temperature.

Turn-around time: Routine: 5-7 working days.

CARBOXYLASE ACTIVITIES **(Lymphocytes, Fibroblasts,)**

(PYRUVATE, PROPIONYL-COA, AND 3-METHYLCROTONYL-COA CARBOXYLASES)

Comments: Radiochemical assays performed on cultured cells or isolated lymphocytes. Carboxylases are routinely run twice a month. Please schedule blood draw on Monday thru Wednesday. Blood for lymphocyte assay must arrive in our laboratory within 36 hours of the sample draw. Samples should be shipped by overnight carrier at room temperature in an insulated container (add freezer gel pack during warmer seasons) to be received at the laboratory by the following morning. Please fill out any relevant patient data on the requisition form to assist in interpretation. **Fibroblast analysis will have an additional charge for culture and storage (see fibroblast/amniocyte culture and storage)**

Sample requirements: Blood, 5.0 mL in ACD (yellow-top) tube (3.0 ml minimum on infants), **Blood for lymphocyte assay must arrive in our laboratory within 36 hours of the sample draw. Samples should be shipped by overnight carrier at room temperature in an insulated container (add freezer gel pack during warmer seasons) to be received at the laboratory by the following morning. Do not heat or freeze. Note: we are closed weekends and holidays.**
Fibroblasts, Two T-25 flasks shipped by overnight carrier at room temperature in an insulated container (add freezer gel pack during warmer seasons) to be received at the laboratory by the following morning.

Turn-around time: Routine: 10-15 working days (not including growing cultured cells, if required).

QUANTITATIVE BENZOIC AND HIPPURIC **(Plasma)**

Comments: May be useful during treatment with benzoate and phenylacetate.

Sample requirements: Plasma, ≥2 mL (minimum 1.0 mL) from heparinized blood (green top tube) supernatant from clinical centrifugation (within 20 minutes) promptly frozen and shipped frozen (packed with dry ice).

Turn-around time: Routine: 5-7 working days.