



# Biochemical Genetics Laboratory

## The Clair Burgener

### Clinical Research Diagnostic Unit

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:**

UCSD Biochemical Genetics Lab  
TGCF Bldg, Room 203  
220 Dickinson St.  
San Diego, CA 92103

**Mailing Address:**

UCSD Biochemical Genetics Lab  
Department of Pediatrics, 0830  
UCSD School of Medicine  
La Jolla CA 92093-0830

Phone: (619) 543-5260  
FAX: (619) 543-3565

**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.
- **We do not bill insurance companies for these services.** Billing must be directed towards the referring laboratory. If the referring laboratory is not responsible for payment, then full payment must accompany specimen before processing. (bank draft or money order made payable to UCSD Biochemical Genetics Laboratory).

Certification numbers:

CLIA ID # O5D0643075                      Federal Tax ID # 33-0833316  
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 clinical research diagnostics unit laboratory.

<b>Test</b>	<b>Specimens</b>
<b>HOLOCARBOXYLASE SYNTHETASE</b>	<b>Blood, Cultured Cells</b>
<b>HYPOXANTHINE-GUANINE PHOSPHORIBOSYL TRANSFERASE FOR CARRIER DETECTION</b>	<b>Hair Roots</b>
<b>HYPOXANTHINE-GUANINE PHOSPHORIBOSYL TRANSFERASE</b>	<b>Cultured Cells</b>
<b>OXPURINE QUANTITATION</b>	<b>Urine</b>

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

## METABOLIC ASSAYS

---

### HYPOXANTHINE-GUANINE PHOSPHORIBOSYL TRANSFERASE FOR CARRIER DETECTION

**(Hair Roots)**

Comments: To determine heterozygosity for female carriers of this X-linked disorder, distribution of HPRT activity can be determined among individual hair root bulbs from the potential carrier. Statistical analysis indicates that analysis of  $\geq 35$  hair roots can exclude carrier status to a certainty of 95%.

Sample requirements: Hair roots, minimum of 35 bulbs in good condition. Storage and shipping at room temp is adequate. Please call our lab for an instruction sheet on collection of hair roots.

Turn-around time: 1-2 weeks

---

### HYPOXANTHINE-GUANINE PHOSPHORIBOSYL TRANSFERASE

**(Cultured Fibroblasts, Amniocytes and Chorionic Villi)**

Comments: The diagnosis of Lesch-Nyhan syndrome and variant forms depends on the assay of HPRT. Our assay is radiochemical, and we also measure Adenine Phosphoribosyl Transferase as an internal control. It is helpful to have a clinical history of the patient provided. **Fibroblast, Amniocyte and Chorionic Villi analysis will have an additional charge for culture (see fibroblast/amniocyte culture).**

Sample requirements: Cultured fibroblasts, 2 T-25 flasks shipped with sterile medium at room temp overnight. **Please call lab to coordinate**

Amniocytes and Chorionic Villi, **Please call lab to coordinate.**

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

---

### OXYPURINE QUANTITATION

**(Urine)**

Comments: The ratio of excretion of the oxypurines (hypoxanthine, xanthine, uric acid) may be used to distinguish among conditions of hypouricemia (renal tubular wasting *vs* xanthine oxidase or molybdenum cofactor deficiency), and to monitor therapy in hyperuricemic conditions (*e.g.* gout and Lesch-Nyhan syndrome). We use an HPLC method to obtain ratios of the oxypurines to creatinine.

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

Turn-around time: 2-8 weeks.

---

### HOLOCARBOXYLASE SYNTHETASE

**(Lymphocytes, Fibroblasts, Amniocytes)**

Comments: This is a radiochemical assay performed on cultured cells or isolated lymphocytes. For assay of lymphocytes from a blood sample, please call to assure that staff is available, as the specimens must be processed stat upon arrival. **Fibroblast/Amniocyte analysis will have an additional charge for culture (see fibroblast/amniocyte culture).**

Sample requirements: Blood, 10 mL in ACD (yellow-top) tube, kept at room temperature and shipped at room temperature overnight. For greater reliability, we **require** a simultaneous sample from a control individual (not a member of the patient's family).

Fibroblasts, Two T-25 flasks shipped overnight with medium at room temperature.

Prenatal diagnosis is possible with Amniocytes: Please call lab to coordinate.

Turn-around time: Routine: 5-10 working days (not including growing cultured cells, if required).

**PLEASE CONTACT LABORATORY TO ARRANGE FOR THIS ASSAY ON LYMPHOCYTES**

---

**TISSUE CULTURE SERVICES****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.

**FIBROBLAST/AMNIOCYTE CONTINUED 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 or amniocytes), 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/AMNIOCYTE 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/AMNIOCYTE 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.