



BIOCHEMICAL GENETICS LABORATORY
 PEDIATRICS — CTF BLDG., ROOM B-213
 UNIVERSITY OF CALIFORNIA, SAN DIEGO
 212 DICKINSON ST. — SAN DIEGO, CA 92103
 (619) 543-5260 — FAX (619) 543-3565
 LAB DIRECTOR: B.A. BARSHOP, M.D.,PH.D.

| | |
|--|----------|
| PATIENT'S NAME | |
| DATE OF BIRTH | SEX: M F |
| ID # | |
| <input type="checkbox"/> INPATIENT <input type="checkbox"/> OUTPATIENT | |

| | | |
|-------------------|-------------------|---------------|
| UCSD LAB USE ONLY | UCSD PATIENT ID # | UCSD SAMPLE # |
|-------------------|-------------------|---------------|

| | | |
|--|-----------------|--------------------|
| PHYSICIAN INFORMATION REQUIRED FOR BILLING PURPOSES | | DATE/TIME RECEIVED |
| REQUESTING PHYSICIAN | STATE LICENSE # | |

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|-----------------|---|
| RESULTS ADDRESS | BILLING ADDRESS (WE DO NOT BILL PATIENTS) |
| TELEPHONE | TELEPHONE |

TEST REQUESTED

| | | | | |
|--|-----------------|------|-------------------|---------------|
| URINE | COLLECTION DATE | TIME | UCSD LAB USE ONLY | UCSD SAMPLE # |
| <input type="checkbox"/> QUALITATIVE METABOLIC TESTS <input type="checkbox"/> METHYLMALONIC ACID <input type="checkbox"/> SUCCINYLACETONE <input type="checkbox"/> QUANTITATIVE AMINO ACIDS <input type="checkbox"/> N-ACETYLASPARTIC ACID <input type="checkbox"/> OROTIC ACID <input type="checkbox"/> QUANTITATIVE ORGANIC ACIDS COMPREHENSIVE <input type="checkbox"/> QUANTITATIVE ORGANIC ACIDS SCREEN (This is a limited assay) <input type="checkbox"/> CARNITINE <input type="checkbox"/> SUCCINYLPURINE SCREEN <input type="checkbox"/> OTHER _____ | | | | |

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|--|-----------------|------|-------------------|---------------|
| PLASMA | COLLECTION DATE | TIME | UCSD LAB USE ONLY | UCSD SAMPLE # |
| <input type="checkbox"/> QUANTITATIVE AMINO ACIDS <input type="checkbox"/> CARNITINE <input type="checkbox"/> QUANTITATIVE ORGANIC ACIDS COMPREHENSIVE <input type="checkbox"/> HOMOCYST(E)INE <input type="checkbox"/> ACYLCARNITINE <input type="checkbox"/> COENZYME Q10 <input type="checkbox"/> PKU PANEL (Phenylalanine, Tyrosine) <input type="checkbox"/> OTHER _____ | | | | |

| | |
|--|--------------------------|
| PLASMA, URINE | COLLECTION DATE AND TIME |
| Please record above in spaces for plasma and urine sample entry | |
| <input type="checkbox"/> METABOLIC PANEL (QUANTITATIVE PLASMA AMINO ACIDS AND QUANTITATIVE URINE ORGANIC ACIDS SCREEN) | |

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|---|-----------------|------|-------------------|---------------|
| CSF | COLLECTION DATE | TIME | UCSD LAB USE ONLY | UCSD SAMPLE # |
| <input type="checkbox"/> QUANTITATIVE AMINO ACIDS <input type="checkbox"/> OTHER _____ <input type="checkbox"/> QUANTITATIVE ORGANIC ACIDS COMPREHENSIVE | | | | |

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|---|-----------------|------|-------------------|---------------|
| WHOLE BLOOD | COLLECTION DATE | TIME | UCSD LAB USE ONLY | UCSD SAMPLE # |
| <input type="checkbox"/> HGPRT (PKU Card) <input type="checkbox"/> PKU PANEL (Phenylalanine, Tyrosine – PKU Card) <input type="checkbox"/> MCAD (Medium Chain Acyl CoA Dehydrogenase Common Allele) <input type="checkbox"/> CARBOXYLASES IN LYMPHOCYTES :pyruvate, propionyl CoA, 3-methylcrotonyl CoA (control required) | | | | |

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|--|-----------------|------|-------------------|---------------|
| OTHER | COLLECTION DATE | TIME | UCSD LAB USE ONLY | UCSD SAMPLE # |
| <input type="checkbox"/> ESTABLISH FIBROBLAST CULTURE <input type="checkbox"/> FIBROBLAST STORAGE <input type="checkbox"/> FIBROBLAST CONTINUED CULTURE <input type="checkbox"/> FIBROBLAST RECULTURED FROM STORAGE <input type="checkbox"/> CARBOXYLASES :pyruvate, propionyl CoA, 3-methylcrotonyl CoA (Specify source: fibroblasts or amniocytes) | | | | |

TO ASSIST IN INTERPRETATION

CLINICAL HISTORY

| CODE | DIAGNOSIS | CODE | DIAGNOSIS | CODE | DIAGNOSIS | CODE | DIAGNOSIS | CODE | DIAGNOSIS |
|--------|---------------------------------|-------|-------------------|-------|----------------|-------|--------------------|--------|------------|
| 276.2 | ACIDOSIS | 369.9 | HEARING LOSS | 271.8 | HYPEROXALURIA | 251.2 | LOW GLUCOSE | 435.9 | STROKES |
| 791.3 | CARNITINE PALMITOYL TRANSFERASE | 746.9 | HEART DISEASE | 251.2 | HYPOGLYCEMIA | 277.9 | METABOLIC DISORDER | 270.6 | UREA CYCLE |
| 780.01 | COMA | 270.6 | HIGH AMMONIA | 781.3 | HYPOTONIA | 270.3 | MSUD | 787.03 | VOMITING |
| 783.4 | DEVELOPMENTAL DELAY | 271.3 | HIGH LACTATE | 791.6 | KETONURIA | 270.1 | PKU | | |
| 250.0 | DIABETES | 277.2 | HPRT; LESCH-NYHAN | 593.9 | KIDNEY DISEASE | 765.1 | PREMATURE | | |
| 783.4 | FAILURE TO THRIVE | 270.6 | HYPERAMMONEMIA | 330.8 | LEIGH DISEASE | 780.3 | SEIZURES | | |

ANY RELEVANT INFORMATION _____

THERAPY & DIET FOR LAST 72 HOURS

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|--|--|---|---|------------------------------------|
| <input type="checkbox"/> X-RAY CONTRAST | <input type="checkbox"/> VALPROIC ACID | <input type="checkbox"/> AMPICILLIN | <input type="checkbox"/> HYPERAL/TPN | <input type="checkbox"/> CARNITINE |
| <input type="checkbox"/> NORMAL DIET FOR AGE | <input type="checkbox"/> PREGESTAMIL | <input type="checkbox"/> BREAST MILK ONLY | <input type="checkbox"/> PREMIE FORMULA | <input type="checkbox"/> MCT OIL |
| <input type="checkbox"/> SPECIAL FORMULAS: | <input type="checkbox"/> OTHER _____ | <input type="checkbox"/> NUTRAMIGEN | | |