

## BIOCHEMICAL GENETICS/CYSTINE LAB

PEDIATRICS, CTFB BLDG., RM. 213

PATIENT NAME:			
DATE OF BIRTH:	SEX:	М	F
PATIENT ID #:			

UNIVERSITY OF CALIFORNIA, SAN DIEGO 212 DICKINSON ST. SAN DIEGO, CA 92103 PH: (619) 543-5260 FAX: (619) 543-3565 LAB DIRECTORS: B.A. BARSHOP, M.D., PhD. AND W.L. NYHAN, M.D., PhD.  CLIA ID: 05D0643075 TAX ID: 33-0833316 NPI ID: 1932264413  UCSD LAB USE ONLY  1. PHYSICIAN INFORMATION REQUIRED FOR BILLING REQUESTING PHYSICIAN: STATE LIC								IENT	F 	
RESULTS ADDRESS				BI	LLING ADDRESS (W	E DO NOT BILL	PATIENTS O	R INSURANCE)		
NAME:				NA	ME:					
HOSPITAL:				HOSPITAL:						
STREET:	CITY:	STATE: ZII	P:	STREET: CITY:				STATE: ZIP:		
PHONE:	PHONE: FAX:			PH	ONE:		FAX:			
2. SAMPLE INFORMATION (	ONLY ONE SPECIME	N TYPE PER RE	QUISITION, PLE	ASI	E)					
COLLECTION DATE:  3. TEST/SERVICE SELECTION		COLLECTION			AM PM	SPECIMEN KE U - URINE P - PLASMA	<b>M</b> - M	USCLE .OODSPOT		
MARK THE TEST OR SERVICE YOU A AT RIGHT). PROVIDE ADDITIONAL	ARE REQUESTING AI			•		C - CSF	<b>F</b> - FIB	ROBLAST <b>BX</b> - SKIN B VHITE BLOOD CELLS	NOPSY	
ORGANIC ACIDS CIRCLE			CIMEN TYPE	A۱	MINO ACIDS			CIRCLE SPECIMEN TYP	PE	
☐ QUANTITATIVE ORGANIC ACIDS U P			□ QU.		QUANTITATIVE AMIN	QUANTITATIVE AMINO ACIDS		U P C		
☐ METHYLMALONIC ACID U P				☐ HOMOCYST(E)INE, TOTAL				Р		
□ N-ACETYLASPARTIC ACID		U		☐ PKU PANEL (PHE/TYR)				BS		
□ OROTIC ACID	OROTIC ACID U			MISCELLANEOUS				CIRCLE SPECIMEN TYPE		
□ SUCCINYLACETONE U				☐ ACYLCARNITINE PROFILE				Р		
CYSTINOSIS TESTING					CARBOXYLASES ENZY	ME ASSAY		B F		
CYSTINOSIS DIAGNOSIS (CYSTIN	NE ASSAY)	B WBC		(PYRUVATE, PROPIONYL COA, 3-METHYLCROTONYL COA)  CLINICAL HISTORY NEEDED FOR INTERPRETATION  (SEE SECTION 4)						
☐ CYSTINOSIS MONITORING (CYSTINE ASSAY) B WB(		B WBC	☐ CARNITINE, TOTAL, FREE		REE		U P M			
SELECT MEDICATION PATIEN	IT IS TAKING			☐ HGPRT ENZYME ASSAY (LESCH-NYHAN DISEASE)			BS			
☐ CYSTAGON ☐ PROCYSBI				□ SUCCINYLPURINE SCREEN				U		
(see www.cystinosiscentral.org f	or sample requirem	ents, shipping	and handling)							
DATE, TIME OF LAST 2 DOSES OF MEDI	CATION (REQUIRED FO	OR ALL CYSTINOS	SIS TESTING)	ВІ	OPSY/CELL CULTUR	RE		CIRCLE SPECIMEN TYP	PE	
DATE: TIME:	A	M PM		☐ ESTABLISH FIBROBLAST CULTURE				BX		
DATE: TIME: AM P			PM		☐ FIBROBLAST CONTINUED CULTURE			F		
					FIBROBLAST STORAGE	Ē		F		
					FIBROBLAST RECULTU	RED FROM STO	DRAGE	F		
4. PROVIDE CLINICAL HISTO	RY TO ASSIST IN	INTERPRET	ATION							
CIRCLE THOSE THAT APPLY		DEVELOPMENTAL DELAY		KETONURIA				PKU		
ABNORMAL NEWBORN SCREENING RESULTS	DIABETES		HPRT; LESCH-NYHAN	IPRT; LESCH-NYHAN KIDNEY		Y DISEASE		SEIZURES		
IDOSIS FATTY ACID OXIDATION DISORDER		HYPERAMMONEMIA			L METABOLISM DISORDER		STROKES			
CARNITINE PALMITOYL TRANSFERASE FAILURE TO THRIVE  COMA HEARING LOSS		HYPEROXALURIA HYPERGLYCEMIA					UREA CYCLE DISORDER VOMITING			
HEART DISEASE/CARDIOMYOPATHY			HYPOTONIA		MSUD					
COMMENTS OR SPECIAL INSTRUC	TIONS:									