

## **BIOCHEMICAL GENETICS/CYSTINE LAB**

PEDIATRICS, CTFB BLDG., RM. 213 UNIVERSITY OF CALIFORNIA, SAN DIEGO

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

PH: (619) 543-5260 FAX: (619) 543-3565  LAB DIRECTORS: B.A. BARSHOP, M.D., PhD.  AND W.L. NYHAN, M.D., PhD.			PATIENT ID #:								
CLIA ID: 05D0643075 TAX ID: 33-08	333316 NPI ID: 1	.932264413	☐ INPATIENT ☐ OUTPATI						IENT		
UCSD LAB USE ONLY LOC CODE:			UCSD SAMPLE #: DATE/TIME						RECEIVED:		
1. PHYSICIAN INFORMATION R	EOUIRED FOR	BILLING PL	IRPOSES								
REQUESTING PHYSICIAN:		STATE LICENS					UPIN #:				
RESULTS ADDRESS	BILLING ADDRESS (WE DO NOT BILL PATIENTS)										
NAME:			NAME:								
HOSPITAL:	HOSPITAL:										
STREET: CITY: STATE: ZIP:			STREET: CITY: STATE: ZIP								
PHONE:	FAX:			PHONE: FAX:							
2. SAMPLE INFORMATION (ONL	V ONE SPECIMEN	TVDE DER RE	OUISITION DIE	ASE)							
COLLECTION DATE:	TONE SI ECIMEN	COLLECTION		AJL			SPECIMEN KE	y.			
						AM PM	U - URINE	<b>M</b> - MUS	SCLE <b>SL</b> - SALIVA		
3. TEST/SERVICE SELECTION							P - PLASMA	BS - BLO	ODSPOT <b>SW</b> - BUCCAL S	WAB	
MARK THE TEST OR SERVICE YOU ARE	REQUESTING ANI	CIRCLE THE	SPECIMEN TYPI	E (SEE S	SPECIME	N KEY	C - CSF	F - FIBRO	OBLAST BX - SKIN BIOI	PSY	
AT RIGHT). PROVIDE ADDITIONAL REQ	UIRED INFORMAT	TION AS REQU	ESTED AND CLII	NICAL F	HISTORY	BELOW.	B - WHOLE BLO	00D <b>wc</b> - WH	TITE BLOOD CELLS		
ORGANIC ACIDS	CIRCLE SPEC	AMINO ACIDS					CIRCLE SPECIMEN TYPE				
□ QUANTITATIVE ORGANIC ACIDS		U P C		☐ QUANTITATIVE AMINO AC			O ACIDS		U P C		
□ METHYLMALONIC ACID		U P		п н	☐ HOMOCYST(E)INE, TOTAL				Р		
□ N-ACETYLASPARTIC ACID		U		□ PI	□ PKU PANEL (PHE/TYR)				BS		
□ OROTIC ACID		U		MISCELLANEOUS					CIRCLE SPECIMEN TY	/PE	
□ SUCCINYLACETONE		U	□ ACYLCARNITINE PROFILE					P			
MOLECULAR DIAGNOSTICS		CIRCLE SPECIMEN TYPE		☐ CARBOXYLASES ENZYME ASSAY					B F		
☐ DNA EXTRACTION		B M SL SW		(PYRUVATE, PROPIONYL CoA, 3-METHYLCROTON				TONYL CoA)			
☐ MCAD COMMON ALLELE (c.985 A—	→ G)	в м		CONTROL REQUIRED (NO CHARGE): MUST NOT BE A							
☐ MITOCHONDRIAL DNA PANEL		B M SL SW		RELATIVE, MUST BE PREPARED WITH  CLINICAL HISTORY (SECTION 4) REQU							
(INCLUDES POINT MUTATIONS AND SOUTHERN BLOT)			☐ CARNITINE, TOTAL, FREE					U P M			
☐ SINGLE POINT MUTATION CHOOSE BELOW		B M SL SW		☐ COENZYME Q10					P M		
☐ MELAS A3243G ☐ MELAS T3271C				☐ HGPRT ENZYME ASSAY (LESCH-NYHAN				DISEASE)	BS		
☐ MERRF A8344G ☐ MERRF T8356C				□ SUCCINYLPURINE SCREEN				,	U		
					CYSTINE DETERMINATION				CIRCLE SPECIMEN TY	/DE	
		в м		☐ STANDARD ASSAY (FOR				YSTEAMINE)	WC	<i>1</i> L	
			CIRCLE SPECIMEN TYPE		DATE, TIME OF LAST CYSTEAMINE ( <b>REC</b>			·	WC		
□ ESTABLISH FIBROBLAST CULTURE		BX		DATE: TIME: AM PM							
☐ FIBROBLAST CONTINUED CULTURE		F		□ DIAGNOSTIC ASSAY (TO DIAGNOSE CYSTINOSIS)				WC			
☐ FIBROBLAST STORAGE		F			CONTROL REQUIRED (NO CHARGE): CONTROL				WC		
		•	NEED NOT BE ETHN								
☐ FIBROBLAST RECULTURED FROM S	RELATIVE; MUST BE PREPARED WITH SAMPLE										
4. PROVIDE CLINICAL HISTORY											
CIRCLE THOSE THAT APPLY 783.4 DEVELOPMENTAL D				VII.4.**	791.6 KETONURIA		ACE		270.1 PKU		
796.6 ABNORMAL NEWBORN SCREENING RESULTS 250.0 DIABETES 276.2 ACIDOSIS 277.85 FATTY ACID OXIDA		277.2 HPRT; LESCH-N ATION DISORDER 270.6 HYPERAMMON					ASE IRIAL METABOLISM DISORDER		780.3 SEIZURES 435.9 STROKES		
791.3 CARNITINE PALMITOYL TRANSFERASE 783.4 FAILURE TO THRIVE									270.6 UREA CYCLE DISORDER		
780.01 COMA 369.9 HEARING LOSS			251.2 HYPERGLYCEMI		330.8 LEIGH DISEA		SE		787.03 VOMITING		
COMMENTS OR SPECIAL INSTRUCTION	RDIOMYOPATHY	781.3 HYPOTONIA			270.3 MSUD						