MEDICAL COLLEGE OF WISCONSIN

SCREENING TESTS FOR DISORDERS OF FATTY ACID OXIDATION

SHIP TO:

William J. Rhead, M.D., Ph.D.
SAMPLE MAILING ADDRESS:
CHW WAREHOUSE
6740 W. Washington St.
West Allis, WI 53214
(414)-266-2979 (office)
(414)-266-2506 (lab)
email: wrhead@mcw.edu

PATIENT AND BILLING INFORMATION

For cultures sent by mail to our laboratory, we greatly prefer to bill the referring hospital or laboratory for these tests. We suggest having the Department of Pathology in your hospital send the sample to us. Preferably, the attached billing information and authorization should be included with fibroblast cultures and clinical and biochemical information.

Patient Information:

Date: _____ DIAGNOSIS:_____

Patient Name: _____

Date of Birth: MONTH_____ DAY_____ YEAR_____ **Male or Female** (circle one)

Your Hospital #: _____

Date of Biopsy: MONTH_____ DAY_____ YEAR_____

Referring Physician Information:

Name: _____

Address: _____

City/State/Zip _____

Phone/Fax: _____

Billing Information:

Institution Address: _____

_____

_____

Phone Number: _____

Fax Number: _____

Billing Authorization

Signature: _____

Genetics Center
Children's Hospital of Wisconsin
9000 W Wisconsin Avenue, MS #716
Milwaukee, WI 53226

MEDICAL COLLEGE OF WISCONSIN

SCREENING TESTS (ICD-10: E78.9) FOR DISORDERS OF FATTY ACID OXIDATION (ICD10-10: E71.31)

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Monday through Thursday -

Ship two T-25 ($25cm^2$) flasks of confluent fibroblasts obtained from skin biopsy and routine fibroblast tissue culture to the above address by overnight express accompanied by clinical and biochemical data and the billing information sheet.

> ^{3}H-Acyl-carnitine profiling by radio-HPLC in intact cultured fibroblast monolayers to screen for all known disorders of β-oxidation, excluding riboflavin-responsive variants and all methylmalonic acidemias: **$998.25** (CPT codes 82017+88233+82542), with physician interpretation (CPT code 80500).

Prenatal Diagnosis

Prenatal diagnosis on amniocytes is possible for virtually all fatty acid oxidation disorders. However, before we will accept amniocyte samples from a referring laboratory, please contact William J. Rhead, MD, PhD directly at 414-266-2906. In addition to 2 confluent T-25 flasks of amniocytes from the proband fetus, we require amniocytes from 2 anonymous control pregnancies, 2 T-25s each. Further details can be obtained directly from Dr. Rhead.

> ^{3}H-Acyl-carnitine profiling by radio-HPLC in amniocytes to screen for all known disorders of β-oxidation, excluding riboflavin-responsive variants and all methylmalonic acidemias: **$998.50** (CPT codes 88235+82542), with physician interpretation (CPT code 80500).

These analyses are non-experimental, non-investigational and not in conflict with medical standards. This laboratory and Children's Hospital of Wisconsin has been accredited by the Joint Commission on Accreditation of Healthcare Organizations, the College of American Pathologists, CLIA (52D0661957), and HCFA (license number 16-0058). Dr. Rhead is board-certified in Clinical Biochemical Genetics by the American Board of Medical Genetics.

EXPLANATION OF CPT CODES:

88233	Skin fibroblast culture
82492	Chromatography, quantitative, column (eg, gas liquid or HPLC); multiple analytes, single stationary and mobile phase
88235	Tissue culture for non-neoplastic disorders; amniotic fluid or chorionic villus cells
80500	Physician interpretation
88372	Protein analysis of tissue by Western Blot, with interpretation and report, one band

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email: wrhead@mcw.edu

Certification:
American Board of Pediatrics - 1981
American Board of Medical Genetics
Clinical Genetics - 1982
Clinical Biochemical Genetics – 1993, 2003, 2005, 2007, 2009

LABORATORY NAME:	Fatty Acid Oxidation
LAP NUMBER .:	1780401
LAST JCAHO ACCREDITATION DATE:	June 2016
CLIA IDENTIFICATION NO.:	52D0661957, expires 01/17
MEDICARE NO.:	160058
MEDI-CAL PROVIDER NO.:	XYP173950
STATE LICENSE NO.:	Wisconsin 42007
FEDERAL TAX ID NO.:	39-0806261N
HCFA LICENSE NO.:	16-0058
UPIN #:	A01404

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MEDICAL COLLEGE OF WISCONSIN

Dear Colleagues: January 2017

This letter will update you concerning the diagnostic services available in the Fatty Acid Oxidation Disorder Laboratory at the Medical College of Wisconsin and Children's Hospital of Wisconsin. For almost 18 years, we have offered these services to geneticists, endocrinologists, gastroenterologists and neurologists around the country. Using radioHPLC, we can now define the characteristic diagnostic profiles of accumulated fatty acyl-carnitines in cultured fibroblasts from affected patients and can also detect isovaleric acidemia, all subtypes of propionic and methylmalonic acidemias and β-ketothiolase deficiency. We have also performed many successful prenatal diagnoses for these disorders in cultured amniocytes. The unique panel of diagnostic tests we offer is summarized below.

My group also collaborates closely with other major North American, European, Japanese and Australasian academic groups concerned with the diagnosis, treatment, and investigation of fatty acid oxidation disorders. I am board certified in Clinical Biochemical Genetics and Clinical Genetics and the laboratory is accredited by CAP, JCAHO and registered under CLIA. Please call, write, e-mail or fax me if you want additional testing information or wish to discuss individual patients.

Sincerely yours,

William J. Rhead, MD PhD
Professor of Pediatrics
E-mail: wrhead@mcw.edu
(414) 266-2906; FAX (414) 266-1616

DIAGNOSTIC TESTS:

Enzyme Deficiency: (Abbreviation)	Diagnostic RadioHPLC Profile:
Acyl-CoA Dehydrogenases (ACD):	
Short-Chain (SCAD); 3-Hydroxy Short Chain (SCHAD)	X
Medium-Chain (MCAD)	X
Very-Long-Chain (VLCAD); Long-Chain (LCAD)	X
3-Hydroxy-Long-Chain (LCHAD)	X
Glutaric Acidemia, Type II (GA-II):	
Electron-Transfer Flavoprotein (ETF)	X
Electron-Transfer Flavoprotein Dehydrogenase (ETF-DH; ETF-QO)	X
Trifunctional Protein (TFP)	X
Carnitine Palmityl Transferases, I and II (CPT-I and -II)	X; X
Carnitine Acyl-Carnitine Translocase (CACT)	X
Carnitine Uptake Defect (CUD); Systemic Carnitine Deficiency	X
Methylmalonic and Propionic Acidemias, ALL subtypes	X
β-Ketothiolase	X
Isovaleryl-CoA-dehydrogenase (IVA)	X

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