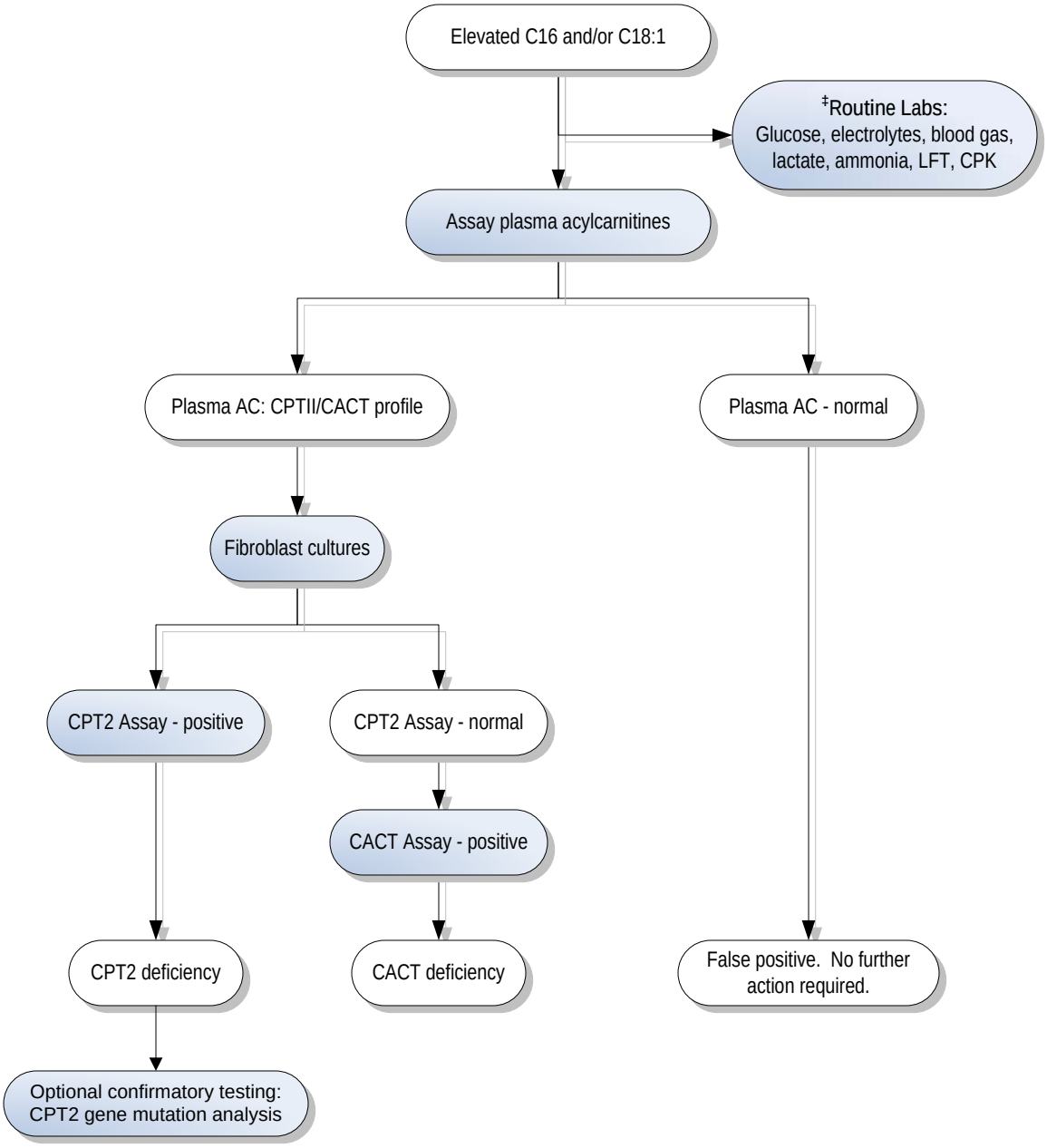
C16 and/or C18:1 Elevated



Abbreviations/Key:
AC = acylcarnitine
CACT = carnitine - acylcarnitine translocase
CPK = creatine phosphokinase
CPT2 = carnitine palmitoyltransferase 2
LFT = liver function tests

‡ = When the positive predictive value of screening is high and the risk to the baby is high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

Actions are shown in shaded boxes; results are in the unshaded boxes.

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

SPECIMEN INFORMATION

Printed By : JDT1303

Specimen # : 20122630554 LastName: JIANHUA
DOC: 09/17/2012 DOR: 09/19/2012 Rptd: 09/21/2012
Name: JIANHUA MR #: 01238373
Mother: CHEN, LIYING Sex: Male
DOB: 09/16/2012 @ 0549 BW: 3316 gms
DOC: 09/17/2012 @ 0810 Twin: Single
AAC: 1 day(s) 2 hour(s) Transfused:
Specimen Age: 2 day(s) Trans Date:
Opt. Use :
Race: Asian/Pacific Islander Ethnic:
HA/TPN: NICU:
Antibiotics: Steroids: 1st screen

Submitter
EVERGREEN HEALTH (H0164)
12040 NE 128TH ST, KIRKLAND, WA 98034
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Physician
JULIE WEN (P44842)
22635 NE MARKET PLACE DR. SUITE 120, REDMOND, WA 9
Phone: (425) 898-7408 Fax: (425) 898-7409

DISORDER	MNEMONIC	STATUS	TEST	VALUE
PISH	NORM	Normal	PISH	9.71
MCADD	NORM	Normal	C8	0.07
			C8/C2	0
			C8/C10	0.73
			C10:1	0.07
PKUMS	NORM	Normal	Phe	65
			Phe/Tyr	0.97
GAL	NORM	Normal	GAL	11.57
BIO	NORM	Normal	BIO	
MSUD	NORM	Normal	Leu	82
			Val	86
			Leu/Ala	0.32
			Leu/Phe	1.27
			Val/Phe	1.34
HCYS	NORM	Normal	Met	23
			Met/Phe	0.36
HGB	FA	Normal	HB-IEF	
CF	NORM	Normal	IRT	8.48
CAH	NORM	Normal	CAH	4.14
ASA	NORM	Normal	CITa	10.52
			ASA	1.05
			Cit/Arg	1.86
			Asa/Arg	0.19
CIT	NORM	Normal	CITc	10.52
TYR-I	NORM	Normal	SUAC	0.31
			Tyr	66.35
HMG	NORM	Normal	C5OHn	0.11
			C5OH/C8	1.46
BKT	NORM	Normal	C5:1	0.01
GA-I	NORM	Normal	C5DC	0.05
			C5DC/C5OH	0.44
			C5DC/C8	0.64
			C5DC/C16	0.02
IVA	NORM	Normal	C5	0.07
			C5/C0	0
			C5/C2	0
			C5/C3	0.06
MMAs/PROP	NORM	Normal	C3	1.15
			C3/C2	0.07
			C3/C16	0.46
MCD	NORM	Normal	C5OHn	0.11
CUD	NORM	Normal	C0	30.21
LCHAD/TFP	NORM	Normal	C16OH	0.03
			C16OH/C16	0.01
VLCAD	NORM	Normal	C14	0.28
			C14:1	0.18
			C16	2.52
			C18	0.91
			C18:1	1.07
			C14:1/C16	0.07

Specimen # : 20122720215 LastName: JIANHUA XIE
DOC: 09/25/2012 DOR: 09/28/2012 Rptd: 10/02/2012
Name: JIANHUA XIE MR #:
Mother: CHEN, LIYING Sex: Male
DOB: 09/16/2012 @ 1749 BW: 3317 gms
DOC: 09/25/2012 @ 1200 Twin: Single
AAC: 8 day(s) 18 hour(s) Transfused:
Specimen Age: 3 day(s) Trans Date:
Opt. Use :
Race: Asian/Pacific Islander Ethnic:
HA/TPN: NICU:
Antibiotics: Steroids: 2nd screen.

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Physician
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DISORDER	MNEMONIC	STATUS	TEST	VALUE
PISH	NORM	Normal	PISH	3.7
MCADD	NORM	Normal	C8	0.08
			C8/C2	0
			C8/C10	0.89
			C10:1	0.09
PKUMS	NORM	Normal	Phe	51
			Phe/Tyr	0.51
GAL	NORM	Normal	GAL	9.47
BIO	NORM	Normal	BIO	
MSUD	NORM	Normal	Leu	184
			Val	168
			Leu/Ala	0.36
			Leu/Phe	3.58
			Val/Phe	3.28
HCYS	NORM	Normal	Met	35
			Met/Phe	0.68
HGB	FA	Normal	HB-IEF	
CF	NORM	Normal	IRT	7.05
CAH	NORM	Normal	CAH	5.43
ASA	NORM	Normal	CITa	16.32
			ASA	2.5
			Cit/Arg	1.09
			Asa/Arg	0.17
CIT	NORM	Normal	CITc	16.32
TYR-I	NORM	Normal	SUAC	0.34
			Tyr	99.52
HMG	NORM	Normal	C5OHn	0.13
			C5OH/C8	1.57
BKT	NORM	Normal	C5:1	0.02
GA-I	NORM	Normal	C5DC	0.03
			C5DC/C5OH	0.2
			C5DC/C8	0.32
			C5DC/C16	0.01
IVA	NORM	Normal	C5	0.16
			C5/C0	0
			C5/C2	0.01
			C5/C3	0.19
MMAs/PROP	NORM	Normal	C3	0.87
			C3/C2	0.05
			C3/C16	0.37
MCD	NORM	Normal	C5OHn	0.13
CUD	NORM	Normal	C0	48.83
LCHAD/TFP	NORM	Normal	C16OH	0.02
			C16OH/C16	0.01
VLCAD	NORM	Normal	C14	0.3
			C14:1	0.14
			C16	2.39
			C18	0.92
			C18:1	1.55
			C14:1/C16	0.06

MAYO CLINIC
Mayo Medical Laboratories

Web: MayoMedicalLaboratories.com
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International: +1 855-379-3115
Values are valid only on day of printing.

Test ID: ACRN
Acylcarnitines, Quantitative, Plasma

Useful For

Diagnosis of fatty acid oxidation disorders and several organic acidurias

Evaluating treatment during follow-up of patients with fatty acid beta-oxidation disorders and several organic acidurias

Testing Algorithm

The following algorithms are available in Special Instructions:
-Newborn Screening Follow-up for Elevations of C8, C6, and C10 Acylcarnitines (also applies to any plasma C8, C6, and C10 acylcarnitine elevations)
-Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations (also applies to any plasma C4 acylcarnitine elevation)
-Newborn Screening Follow-up for Isolated C5 Acylcarnitine Elevations (also applies to any plasma C5 acylcarnitine elevation)

Clinical Information

Acylcarnitine analysis enables the diagnosis of many disorders of fatty acid oxidation and several organic acidurias, as relevant enzyme deficiencies cause the accumulation of specific acyl-CoAs. Fatty acid oxidation (FAO) plays a major role in energy production during periods of fasting. When the body's supply of glucose is depleted, fatty acids are mobilized from adipose tissue, taken up by the liver and muscles, and oxidized to acetyl-CoA. In the liver, acetyl-CoA is the building block for the synthesis of ketone bodies, which enter the blood stream and provide an alternative substrate for production of energy in other tissues when the supply of glucose is insufficient to maintain a normal level of energy. The acyl groups are conjugated with carnitine to form acylcarnitines, which are measured by tandem mass spectrometry (MS/MS). Diagnostic results are usually characterized by a pattern of significantly elevated acylcarnitine species compared to normal and disease controls.

In general, more than 20 inborn errors of metabolism can be identified using this method including FAO disorders and organic acidurias. The major clinical manifestations associated with individual FAO disorders include hypoketotic hypoglycemia, variable degrees of liver disease and failure, skeletal myopathy, dilated/hypertrophic cardiomyopathy, and sudden or unexpected death. Organic acidurias also present as acute life-threatening events early in life with metabolic acidosis, increased anion gap, and neurologic distress. Patients with any of these disorders are at risk of developing fatal metabolic decompensations following the acquisition of even common infections. Once diagnosed, these disorders can be treated by avoidance of fasting, special diets, and cofactor and vitamin supplementation.

Analysis of acylcarnitines in blood and bile spots represents the first level of evaluation of a complete postmortem investigation of a sudden or unexpected death of an individual. Additional confirmatory testing is recommended. The diagnosis of an underlying FAO disorder or organic aciduria allows genetic counseling of the family, including the possible option of future prenatal diagnosis, and testing of at-risk family members of any age.

Disorders Detectable by Acylcarnitine Analysis*

Fatty Acid Oxidation Disorders:

-Carnitine palmitoyltransferase I (CPTI) deficiency
-Medium-chain 3-ketoacyl-CoA thiolase (MCKAT) deficiency
-Dienoyl-CoA reductase deficiency
-Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
-Medium/Short-chain 3-hydroxyacyl-CoA dehydrogenase (M/SCHAD) deficiency
-Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
-Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency & trifunctional protein deficiency
-Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
-Carnitine palmitoyl transferase type II (CPT-II) deficiency
-Carnitine-acylcarnitine translocase (CACT) deficiency
-Electron transfer flavoprotein (ETF) deficiency, ETF-dehydrogenase deficiency (multiple acyl-CoA dehydrogenase deficiency [MADD]; glutaric acidemia type II)

Organic Acid Disorders:

-Glutaryl-CoA dehydrogenase deficiency (glutaric acidemia type I)
-Propionic Acidemia
-Methylmalonic Acidemia
-Isovaleric Acidemia
-3-hydroxy-3-methylglutaryl-CoA carboxylase deficiency
-3-Methylcrotonyl carboxylase deficiency
-Biotinidase deficiency
-Multiple carboxylase deficiency
-Isobutyryl-CoA dehydrogenase deficiency
-2-Methylbutyryl-CoA dehydrogenase deficiency
-Beta-ketothiolase deficiency
-Malonic aciduria
-Ethylmalonic encephalopathy
-Glutamate formiminotransferase deficiency (Formiminoglutamic aciduria)

*Further confirmatory testing is required for most of these conditions because an acylcarnitine profile can be suggestive of more than 1 condition.

Reference Values

	< or =7 days (nmol/mL)	8 days-7 years (nmol/mL)	> or =8 years (nmol/mL)
Acetylcarnitine, C2	2.14-15.89	2.00-27.57	2.00-17.83
Acrylylcarnitine, C3:1	<0.04	<0.05	<0.07
Propionylcarnitine, C3	<0.55	<1.78	<0.88
Formiminoglutamate, FIGLU	<0.43	<0.08	<0.14
Iso-/Butyrylcarnitine, C4	<0.46	<1.06	<0.83
Tiglylcarnitine, C5:1	<0.05	<0.09	<0.11
Isovaleryl-/2-Methylbutyrylcarn C5	<0.38	<0.63	<0.51
3-OH-iso-/butyrylcarnitine, C4-OH	<0.13	<0.51	<0.18
Hexenoylcarnitine, C6:1	<0.12	<0.10	<0.15
Hexanoylcarnitine, C6	<0.14	<0.23	<0.17
3-OH-isovalerylcarnitine, C5-OH	<0.08	<0.12	<0.10
Benzoylcarnitine	<0.13	<0.07	<0.10
Heptanoylcarnitine, C7	<0.05	<0.05	<0.06
3-OH-hexanoylcarnitine, C6-OH	<0.08	<0.19	<0.09
Phenylacetylcarnitine	<0.15	<0.22	<0.29
Salicylcarnitine	<0.08	<0.09	<0.09
Octenoylcarnitine, C8:1	<0.48	<0.91	<0.88
Octanoylcarnitine, C8	<0.19	<0.45	<0.78
Malonylcarnitine, C3-DC	<0.09	<0.14	<0.26
Decadienoylcarnitine, C10:2	<0.11	<0.12	<0.26
Decenoylcarnitine, C10:1	<0.25	<0.46	<0.47
Decanoylcarnitine, C10	<0.27	<0.91	<0.88
Methylmalonyl-/succinylcarn, C4-DC	<0.05	<0.05	<0.05
3-OH-decenoylcarnitine, C10:1-OH	<0.12	<0.12	<0.13
Glutarylcarnitine, C5-DC	<0.06	<0.10	<0.11
Dodecenoylcarnitine, C12:1	<0.19	<0.37	<0.35
Dodecanoylcarnitine, C12	<0.18	<0.35	<0.26
3-Methylglutarylcarnitine, C6-DC	<0.28	<0.21	<0.43
3-OH-dodecenoylcarnitine, C12:1-OH	<0.11	<0.10	<0.13
3-OH-dodecanoylcarnitine, C12-OH	<0.06	<0.09	<0.08
Tetradecadienoylcarnitine, C14:2	<0.09	<0.13	<0.18
Tetradecenoylcarnitine, C14:1	<0.16	<0.35	<0.24
Tetradecanoylcarnitine, C14	<0.11	<0.15	<0.12
Octanedioylcarnitine, C8-DC	<0.25	<0.19	<0.19
3-OH-tetradecenoylcarnitine C14:1OH	<0.06	<0.18	<0.13
3-OH-tetradecanoylcarnitine, C14-OH	<0.04	<0.05	<0.08
Hexadecenoylcarnitine, C16:1	<0.15	<0.21	<0.10
Hexadecanoylcarnitine, C16	<0.36	<0.52	<0.23
3-OH-hexadecenoylcarnitine,C16:1-OH	<0.78	<0.36	<0.06
3-OH-hexadecanoylcarnitine, C16-OH	<0.10	<0.07	<0.06
Octadecadienoylcarnitine, C18:2	<0.12	<0.31	<0.24
Octadecenoylcarnitine, C18:1	<0.25	<0.45	<0.39
Octadecanoylcarnitine, C18	<0.10	<0.12	<0.14
Dodecanedioylcarnitine, C12-DC	<0.10	<0.04	<0.04
3-OH-octadecadienoylcarn, C18:2-OH	<0.04	<0.06	<0.06
3-OH-octadecenoylcarnitine C18:1-OH	<0.03	<0.04	<0.06
3-OH-octadecanoylcarnitine, C18-OH	<0.03	<0.05	<0.03

Interpretation

An interpretive report is provided. The individual quantitative results support the interpretation of the acylcarnitine profile but are not diagnostic by themselves. The interpretation is based on pattern recognition.

Abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease. To verify a preliminary diagnosis based on an acylcarnitine analysis, independent biochemical (eg, in vitro enzyme assay) or molecular genetic analyses are required.

For information on the follow-up of specific acylcarnitine elevations, see Special Instructions for the following algorithms:
-Newborn Screening Follow-up for Elevations of C8, C6, and C10 Acylcarnitines (also applies to any plasma C8, C6, and C10 acylcarnitine elevations)
-Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations (also applies to any plasma C4 acylcarnitine elevation)
-Newborn Screening Follow-up for Isolated C5 Acylcarnitine Elevations (also applies to any plasma C5 acylcarnitine elevation)

Cautions

In a few instances, false-negative results occur in the analysis of acylcarnitine profiles. For some disorders, such as medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, the calculation of ratios between different acylcarnitine species provides a discriminate factor to overcome such problems. Where applicable, the calculation of such ratios will be incorporated in the routine acylcarnitine analysis. Informative profiles may also not be detected in some disorders where the accumulation of diagnostic acylcarnitines is a reflection of the residual activity of the defective enzyme, the dietary load of precursors, and the anabolic/catabolic and treatment status of a patient.

Patients with carnitine deficiency may not exhibit abnormally high acylcarnitine concentrations. If the results are indicative for carnitine deficiency, the interpretation will include a remark that this limits the diagnostic value of the test and repeat analysis may be considered following carnitine supplementation.

Follow-up testing such as in vitro enzyme assays or molecular genetic testing may be recommended following abnormal acylcarnitine results. It is not advisable to intentionally stress the patient's metabolism (eg, fasting test) prior to specimen collection for acylcarnitine analysis.

Clinical Reference

1. Matern D: Acylcarnitines, including in vitro loading tests. In Laboratory Guide to the Methods in Biochemical Genetics. Edited by N Blau, M Duran, KM Gibson. Springer Verlag 2008 pp 171-206
2. Rinaldo P, Cowan TM, Matern D: Acylcarnitine profile analysis. Genet Med 2008;10:151-156
3. Smith EH, Matern D: Acylcarnitine analysis by tandem mass spectrometry. Curr Protoc Hum Genet 2010;Chapter 17:Unit 17.8.1-20

Special Instructions and Forms

- Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations (also applies to any plasma C4 acylcarnitine elevations)
- Newborn Screening Follow-up for Elevations of C8, C6, and C10 Acylcarnitine (also applies to any plasma C8, C6, and C10 acylcarnitine elevations)
- Newborn Screening Follow-up for Isolated C5 Acylcarnitines Elevations (also applies to any plasma C5 acylcarnitine elevation)

Tell Us What You Think

Seattle Children's Hospital
206-987-2000

Acylcarnitine Profile, Blood

XIE, JIANHUA DRACO - 1275567

* Final Report *

Result Type:	Acylcarnitine Profile, Blood
Result Date:	12 April 2013 10:25
Result Status:	Auth (Verified)
Performed By:	Jack, Rhona , PhD on 17 April 2013 14:09
Verified By:	Jack, Rhona , PhD on 17 April 2013 14:09
Encounter info:	94243980, CHMC, Inpatient, 4/9/2013 - 4/25/2013

* Final Report *

Acylcarnitine Profile/Tandem Mass Spectrometry

Acylcarnitine	Conc mcmole/l	Ref Range >1mo	Acylcarnitine	Conc mcmole/l		Ref Range >1mo
free carnitine	17.11	16-65	C-12:1	0.11		0-0.18
C-2 (acetyl)	22.67	2-24	C-12	0.15		0-0.17
C-3 (propionyl)	0.18	0-0.9	C-6-dicarboxylic	0.04		0-0.08
C-4 (butyryl)	0.13	0-0.7	C-14:2	0.05		0-0.11
C-5:1	0.01	0-0.08	C-14:1	0.10		0-0.18
C-5	0.06	0-0.38	C-14	0.11		0-0.12
C-6	0.09	0-0.23	C-14-hydroxy	0.02		0-0.04
C-5-hydroxy	0.02	0-0.14	C-16:1	0.10	H	0-0.08
C-7	0.02	0-0.06	C-16	0.26	H	0-0.24
C-8:1	0.17	0-0.66	C-16:1-hydroxy	0.02		0-0.03
C-8 (octanoic)	0.21	0-0.27	C-16-hydroxy	0.02		0-0.03
C-3-dicarboxylic	0.09	0-0.15	C-18:2	0.11		0-0.17
C-10:2	0.03	0-0.08	C-18:1	0.26		0-0.33
C-10:1	0.16	0-0.33	C-18	0.05		0-0.15
C-10	0.22	0-0.36	C-18:2-hydroxy	0.01		0-0.03
C-4-dicarboxylic	0.02	0-0.1	C-18:1-hydroxy	0.01		0-0.04
C-5-dicarboxylic	0.1	0-0.11	C-18-hydroxy	0.01		0-0.03

Testing done in plasma
Interpretation: The mild elevation of C16 and C16:1 is non-specific. Repeat is recommended if clinically indicated.

Rhona Jack, Ph.D.
Biochemical Genetics Laboratory

Completed Action List:
* Order by Chen, Laura Peihan, MD on 12 April 2013 9:29
* VERIFY by Jack, Rhona , PhD on 17 April 2013 14:09