U.S. Department of Health and Human Services

Office of the National Coordinator for Health Information Technology

Newborn Screening
Detailed Use Case
Coding and Terminology Guide
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Table of Contents

Preface

The Personalized Healthcare Workgroup developed a Newborn Screening Coding and Terminology Guide of condition and analyte terminology, codes, and mapping that is provided as a supplement to the use case for the purpose of facilitating development of electronic laboratory reports for newborn screening. Newborn screening is different form other laboratory testing in that results are often reported as screening positive or negative for a condition rather than reporting the quantitative results of the actual test performed. One of the newborn screening recommendations that was approved by the AHIC was to report both the clinical conditions identified and the quantitative analytes measured on the electronic reports so that they can support both patient focused care and population health activities. The attached matrix also bring together a variety of coding systems that may be required for rare disorders genetic disorders and provides LOINC codes to assist in identifying results included in a newborn screening report and documenting the methods used in the laboratory. Newborn dried blood spot screening is usually ordered as a single test or panel and the conditions screened for and the analytes that are measured and the methods of measurement may vary from state to state and over time.

This guide can be used to clearly document and encode the reports to identify the conditions screened for or identified and the test results that are associated those conditions. Maintenance of the newborn screening guide and codes will be an on-going activity as the field of newborn screening changes. The guide will also be used to store genotype information that is associated with specific phenotypes are identified by screening. This activity is just beginning and is not yet included in this version of the matrix, but it will become more important as direct genotype measures are among the results measured by newborn screening tests.

The reports that follow are a work in progress that will continue to evolve with additional input from programs and laboratories that perform newborn screening tests. These reports illustrate the types of coding and terminology that will be available for use in electronic newborn screening reports developed using the harmonized standards that will be selected for implementing the use case. Use of a standard framework for coding and terminology will assist in the comparison of data from different laboratories and help identify gaps in coding that should be addressed before laboratories begin to transmit electronic newborn screening reports. The current set of reports address only the initial screening tests carried out on newborn dried blood spots and by early hearing detection and intervention programs.

Many of the reports refer to ACMG primary, secondary, and other conditions that were defined in a report, Toward a Uniform Screening Panel and System[1], that identified 29 conditions for which screening should be mandated based on criteria scores and evaluation by experts. Additional

conditions were identified for which the cost-effectiveness was less clear as well as other conditions that have a role in the differential diagnosis of a condition in the core panel.

The large number of synonyms for newborn screening conditions reflect the evolving state of scientific knowledge resulting in some conditions named by their clinical syndrome, enzyme deficiency, abnormal analyte measured, or specific genome alterations. The paper, Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels[2] helps to elucidate this process and facilitated development of this guide. The use of ACMG codes has provided an effective unifying framework because they have been used as the basis for decisions on which tests to include in screening programs, and are the basis for the National Newborn Screening Information System (NNSIS) and status reports of the National Newborn Screening and Genetics Resource Center (NNSGRC) that have been harmonized[3] in this guide. Clinical coding systems such as SNOMED and ICD10 do not always provide the necessary granularity in this rapidly evolving field and the National Library of Medicine's Unified Medical Language System (UMLS) is expected to assist this process of on-going mapping of variant terminologies and codes used in newborn screening.

[1]Watson MS, Mann MY, Lloyd-Puryear MA, Rinaldo P, Howell RR [editors]. (2006) Newborn screening: Toward a uniform screening panel and system [Executive summary]. Genet Med 8(Supplement):1S-11S.

[2]Sweetman L, Millington DS, Therrell BL, Hannon WH, Popovich B, Watson MS, Mann MY, Michele A. Lloyd-Puryear MA, van Dyck PC. Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels. Pediatrics 2006;117;308-314.

[3]http://genes-r-us.uthscsa.edu/

Conditions

The Conditions report lists the conditions that can be identified by newborn screening tests along with appropriate diagnostic codes. The report includes conditions that are tested for by tandem mass spectrometry (MS/MS) on newborn dried blood spots, and conditions indentified by non-tandem mass spectrometry tests performed on newborn dried blood spots, as well as hearing loss detected through early hearing detection and intervention (EHDI) programs. Specific genetic causes of hearing loss and further classification of types of hearing loss are described in separate reports as this information is not obtained as a result of the newborn screening tests and this report is limited to initial newborn screening tests.

TBD - 'To be determined' in reference to ICD-10 codes highlights those conditions that still need to be classified.

N/A - 'Non applicable'

EHDI: Congenital Hearing Loss: Hearing Loss

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Hearing Loss	HEAR	N/A	N/A	15188001	H91.93

MS/MS: ACMG Primary Targets: Amino Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Argininosuccinic aciduria [Argininosuccinate lyase]	ASA	207900	4.3.2.1	41013004	E72.22
Citrullinemia type I [Argininosuccinate synthetase]	CIT I	215700	6.3.4.5	398680004	E72.21
Homocystinuria [Cystathionine beta-synthase]	HCY	236200	4.2.1.22	11282001	E72.11
Maple syrup urine disease [Branched-chain alpha-keto acid dehydrogenase]	MSUD	248600	1.2.4.4	27718001	E71.0
Phenylketonuria [Phenylalanine hydroxylase]	PKU	261600	1.14.16.1	7573000	E70.0
Tyrosinemia type I [Fumarylacetoacetate hydrolase]	TYR I	276700	3.7.1.2	410056006	E70.21

MS/MS: ACMG Primary Targets: Fatty Acid Oxidase

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Carnitine uptake defect [Plasma membrane carnitine transporter]	CUD	212140		21764004	TBD

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency [Long-chain L-3-Hydroxy dehydrogenase]	LCHAD	609016	1.1.1.211	237999008	E71.310
Medium-chain acyl-CoA dehydrogenase deficiency [Medium-chain acyl-CoA dehydrogenase]	MCAD	607008	1.3.99.3	128596003	E71.311
Trifunctional protein deficiency [Trifunctional protein (alpha, beta subunit)]	TFP	609015	1.1.1.211	237999008	TBD
Very long-chain acyl-CoA dehydrogenase deficiency [Very long-chain acyl-CoA dehydrogenase]	VLCAD	201475	1.3.99.13	237997005	E71.310

MS/MS: ACMG Primary Targets: Organic Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
3-Hydroxy-3-methylglutaric aciduria [3-Hydroxy-3-methylglutaryl-CoA lyase]	HMG	300438	4.1.3.4	124611007	TBD
3-Methylcrotonyl-CoA carboxylase deficiency [3-Methylcrotonyl-CoA carboxylase (alpha, beta subunit)]	3MCC	210200	6.4.1.4	13144005	TBD
beta-Ketothiolase deficiency [beta-ketothiolase]	BKT	203750	2.3.1.16, 2.3.1.9	124265004	TBD
Glutaric acidemia type I [Glutaryl-CoA deydrogenase]	GA I	231670	1.3.99.7	76175005	E72.3
Isovaleric acidemia [Isovaleryl-CoA dehydrogenase]	IVA	243500	1.3.99.10	87827003	E71.110
Methylmalonic acidemia [Methylmalonyl-CoA mutase]	MUT		5.4.99.2	42393006	E71.120
Methylmalonic acidemia [Adenosylcobalamin synthesis]	CBL A	251100	5.4.99.2	73843004	E71.120
Methylmalonic acidemia [Adenosylcobalamin synthesis]	CBL B	251110	5.4.99.2	82245003	E71.120
Multiple carboxylase deficiency [Holocarboxylase synthetase]	MCD	253270	6.3.4.11	15307001	D81.81
Propionic acidemia [Propionyl-CoA carboxylase]	PROP	606054	6.4.1.3	69080001	E71.121

MS/MS: ACMG Secondary Conditions: Amino Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Argininemia [Arginase]	ARG	207800	3.5.3.1	23501004	E72.21
Citrullinemia type II [Aspartate glutamate carrier (citrin)]	CIT II	605814		30529005	E72.23

Disorders of biopterin biosynthesis [6-Pyruvoyltetrahydropterin synthase]	BIOPT-BIO	261640	4.2.3.12		TBD
Disorders of biopterin regeneration [Dihydropteridine reductase]	BIOPT-REG	261630	1.5.1.34	58256000	TBD
Hypermethioninemia [Methionine adenosyltransferase]	MET	250850	2.5.1.6	37695001	E72.1
Hyperphenylalaninemia (variant, benign) [Phenylalanine hydroxylase]	H-PHE		1.14.16.1	68528007	E70.1
Tyrosinemia type II [Tyrosine transaminase]	TYR II	276600	2.6.1.5	4887000	E70.21
Tyrosinemia type III [4-Hydroxyphenylpyruvate acid oxidase]	TYR III	276710	1.13.11.27	415764005	E70.21

MS/MS: ACMG Secondary Conditions: Fatty Acid Oxidase

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
2,4-Dienoyl-CoA reductase deficiency [2,4-Dienoyl-CoA reductase]	DE RED	222745	1.3.1.34		TBD
Carnitine palmitoyltransferase I deficiency [Carnitine palmitoyltransferase Ia]	CPT I	255120	2.3.1.21	238001003	E71.314
Carnitine palmitoyltransferase II deficiency [Carnitine palmitoyltransferase II]	CPT II		2.3.1.21	124265004	E71.314
Carnitine-acylcarnitine translocase deficiency [Carnitine acylcarnitine translocase]	CACT	255110	2.3.1.21	238003000	E71.312
Glutaric acidemia type II [Electron transfer flavoprotein [ETF] (alpha, beta subunit)]	GA II	608053	1.5.5.1	22886006	E71.313
Medium-chain ketoacyl-CoA thiolase deficiency [Medium-chain ketoacyl-CoA thiolase]	MCKAT	602199	2.3.1.16		TBD
Short-chain acyl-CoA dehydrogenase deficiency [Short-chain acyl-CoA dehydrogenase]	SCAD	201470	1.3.99.2	124166007	E71.312
Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency [Short-chain L-3-hydroxy acyl-CoA dehydrogenase]	SCHAD	601609	1.1.1.35	237998000	E71.312

MS/MS: ACMG Secondary Conditions: Organic Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
2-Methyl-3-hydroxybutyric aciduria [2-Methyl-3-hydroxybutyryl-CoA dehydrogenase]	2M3HBA	300438	1.1.1.178		TBD
2-Methylbutyrylglycinuria [2-Methylbutyryl-CoA dehydrogenase]	2MBG	610006	1.3.99.12		TBD

3-Methylglutaconic aciduria [3-Methylglutaconyl-CoA hydratase]	3MGA	250950	4.2.18	237950009	E71.111
Isobutyrylglycinuria [Isobutyryl-CoA dehydrogenase]	IBD	604773	1.1.1.157		TBD
Malonic acidemia [Malonyl-CoA decarboxylase]	MAL	248360	4.1.1.9	361203007	TBD
Methylmalonic aciduria and homocystinuria [MMA mutase and homocysteine: MTHF methyl transferase]	CBL C	277400	5.4.99.2	74653006	E71.1
Methylmalonic aciduria and homocystinuria [MMADHC protein]	CBL D	277410		31220004	TBD

MS/MS: Other Conditions: Amino Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Carbamoyltransferase deficiency [Carbamoyltransferase I]	CPS	237300		124380007	TBD
Girate atrophy of the retina [Ornithine aminotransferase]	Hyper ORN	258870		314467007	H31.23
Histidinemia [Histidine ammonia-lyase]	HIS	235800		410058007	E70.41
Homocystinuria-megaloblastic anemia [Methyltetrahydrofolate homocysteine methyltransferase]	CBL G	250940		360373000	E72.11
Hydroxyprolinemia [4-Hydroxy L-proline oxidase]	OH PRO	237000		25739007	E72.59
Hyperlysinemia [Lysine:alpha-ketoglutarate reductase]	Hyper LYS	238700		58558003	E72.3
Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome [Ornithine translocase]	HHH	238970		30287008	TBD
Methylcobalamin deficiency [Methylcobalamin]	CBL E	236270		4409006	TBD
Methylene tetrahydrofolate reductase deficiency [5,10-methylene tetrahydrofolate reductase]	MTHFR	607093		79514008	E72.12
Nonketotic hyperglycinemia (glycine encephalopathy) [Glycine cleavage system H protein]	NKHG	605899		237939006	E72.51
Ornithine transcarbamylase deficiency [Ornithine transcarbamylase]	OTC	300461		80908008	E72.4
Pyroglutamic acidemia [Glutathione synthetase]	OXO PRO	266130		39112005	TBD
Pyruvate carboxylase deficiency [Pyruvate carboxylase]	PC	266150		87694001	E74.4

Valinemia [Valine transaminase]	Hyper VAL	277100		47719001	E71.19

MS/MS: Other Conditions: Fatty Acid Oxidase

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Maternal carnitine uptake defect [Plasma membrane carnitine transporter]	CUD (mat)	212140		21764004	TBD

MS/MS: Other Conditions: Organic Acids

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Ethylmalonic encephalopathy [Unknown]	EE	602473	1.5.5.1	193051008	TBD
Formiminoglutamic acidemia [Glutamate formiminotransferase]	FIGLU	229100			TBD
Maternal 3-Methylcrotonyl-CoA carboxylase deficiency [3-Methylcrotonyl-CoA carboxylase (alpha, beta subunit)]	3MCC (mat)		6.4.1.4	13144005	TBD
Maternal glutaric acidemia type I [Glutaryl-CoA deydrogenase]	GA I (mat)	231670	1.3.99.7	76175005	TBD
Primary lactic acidemia (various types) [various enzymes]	LACTIC			190882007	E87.2
Succinyl-CoA ligase deficiency [Succinyl-CoA ligase, beta-subunit]	SUCLA2	603921		83792009	TBD

Non-MS/MS: Cystic Fibrosis:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Cystic fibrosis	CF	602421	N/A	190905008	E84.9

Non-MS/MS: Endocrine Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Congenital Adrenal Hyperplasia (non-classical) [Steroid 21-hydroxylase deficiency]	CAH	201910	1.14.99.10	237752007	E25.0
Congenital Adrenal Hyperplasia (salt-wasting) [Steroid 21-hydroxylase deficiency]	CAH	201910	1.14.99.10	71578002	E25.0
Congenital Adrenal Hyperplasia (simple virilizing) [Steroid 21-hydroxylase deficiency]	CAH	201910	1.14.99.10	52604008	E25.0

Congential Adrenal Hyperplasia [Steroid 11-beta hydroxylase deficiency]	CAH	202010	1.14.15.4	237751000	E25.0

Non-MS/MS: Endocrine Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Congenital Hypothyroidism	CH	N/A	N/A	217710005	E03.1
Secondary Congenital Hypothyroidism	20CH	N/A	N/A	267376007	E03.1
Thyroid-Binding Globulin Deficiency	TBG	314200	N/A	237544006	E07.89

Non-MS/MS: Hemoglobin Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Hb C Beta-thalassemia	C/Beta-thal	N/A	N/A		TBD
Hb D Beta-thalassemia	D/Beta-thal	N/A	N/A		TBD
Hb E Beta-thalassemia	E/Beta-thal	N/A	N/A		TBD
Hb H Disease	Hb H	N/A	N/A	48553001	TBD
Hb S Other	N/A	N/A	N/A		TBD
Hemoglobin Disease Other	N/A	N/A	N/A		TBD
Homozygous Beta-thalassemia	F only	N/A	N/A	26682008	D56.2
Homozygous C Disease	FC	N/A	N/A	51053007	D57.2
Homozygous E Disease	FE	N/A	N/A	25065001	D57.8
S/Beta-thalassemia	S/Beta-thal	N/A	N/A	79592006	D57.4
Sickle C-Disease	S/C	N/A	N/A	35434009	D57.2
Sickle cell anemia	S/S	N/A	N/A	191195005	D57.1

Sickle D Disease	S/D	N/A	N/A	25472008	D58.2
Sickle E Disease	S/E	N/A	N/A	47024008	D58.2
Sickle O-Arab Disease	S/O Arab	N/A	N/A	127048005	D58.2

Non-MS/MS: Hemoglobin Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
FA + Other	N/A	N/A	N/A		TBD
FAC	N/A	N/A	N/A	76050008	TBD
FAD/FAG	N/A	N/A	N/A		TBD
FAE	N/A	N/A	N/A	46248003	TBD
FAS	N/A	N/A	N/A		TBD

Non-MS/MS: Infectious Diseases:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Congenital toxoplasmosis	TOXO	N/A	N/A	73893000	P37.1
Human immunodeficiency virus	HIV	N/A	N/A	187438009	B20-B24

Non-MS/MS: Other Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Biotinidase Deficiency 2	BIO	609019	N/A	8808004	D81.810

Non-MS/MS: Other Disorders:

Condition [Enzyme]	ACMG Code	MIM Code	ENZYME Code	SNOMED Code	ICD10 Code
Classical galactosemia (galactose-1-phosphate uridyltransferase deficiency)	GALT	230400	2.7.7.12	398664009	E74.21

Galactoepimerase deficiency (uridine diphosphate galactose 4-epimerase deficiency)	GALE	230350	5.1.3.2	8849004	E74.2
Galactokinase deficiency	GALK	230200	2.7.1.6	124302001	E74.29

Analytes

The Analytes report lists the analytes or chemical entities that are measured by newborn screening tests along with appropriate LOINC codes that are used to identify specific laboratory result fields on electronic laboratory reports. The report includes conditions that are tested for by tandem mass spectrometry (MS/MS) on newborn dried blood spots, and conditions indentified by non-tandem mass spectrometry tests performed on newborn dried blood spots, as well as hearing loss detected through early hearing detection and intervention (EHDI) programs. This report is limited to the initial newborn screening tests and does include additional measures used for confirmatory testing. Because LOINC codes will have separate values based on methods of testing or units of reporting, there may be more than one analyte entry for the same type of measurement. Analytes also include entries for computed sums and ratios when these values are included on the laboratory reports and have been assigned their own LOINC code to identify the result field. To assist in the use of this report, the MS/MS Analytes are listed in alphabetical order within categories and are also listed in molecular weight order which is the order in which they appear on the laboratory instrumentation. Computed sums and ratios are listed separately and also listed following the primary measurement on which they are based.

TBD - 'To be determined' in reference to LOINC codes highlights those analytes for which LOINC codes are being discussed or under development.

EHDI: Hearing Loss

Analyte	Short Name	LOINC Code	Units
Auditory evoked potentials for screening	AEP	TBD	Pass or Refer
Evoked otoacoustic emissions for screening	EOE	TBD	Pass or Refer

MS/MS: Amino Acids

Analyte	Short Name	LOINC Code	Units
Arginine	ARG	47562-4	µmol/L
Argininosuccinate	ASA	53062-6	µmol/L
Aspartate	ASP	47573-1	µmol/L
Citrulline	CIT	42892-0	µmol/L
Glutamate	GLU	47623-4	µmol/L
Glycine	GLY	47633-3	µmol/L
Histidine	HIS	47643-2	µmol/L

Homocitrulline	HOMOCIT	53158-2	µmol/L
Lysine	LYS	47689-5	µmol/L
Methionine	MET	47700-0	µmol/L
Methylhistidine	CH3HIS	47539-2	µmol/L
Phenylalanine	PHE	29573-3	µmol/L
Proline	PRO	47732-3	µmol/L
Serine	SER	47742-2	µmol/L
Succinylacetone	SUAC	53231-7	µmol/L
Threonine	THR	47784-4	µmol/L
Tryptophan	TRP	53159-0	µmol/L
Tyrosine	TYR	35571-9	µmol/L
Valine	VAL	47799-2	µmol/L

MS/MS: Amino Acids Computed Sums and Ratios

Analyte	Short Name	LOINC Code	Units
Alanine + Beta Alanine + Sarcosine	ALA + BALA + SARC	53150-9	µmol/L
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline	AILE + ILE + LEU + OHPRO	53152-5	µmol/L
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio	[AILE + ILE + LEU + OHPRO] / PHE	53153-3	molar ratio
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine	[AILE + ILE + LEU + OHPRO] / ALA	53154-1	molar ratio
Arginine / Phenylalanine Ratio	ARG / PHE	53398-4	TBD
Argininosuccinate / Arginine Ratio	ASA / ARG	53200-2	molar ratio
Asparagine + Ornithine	ASN + ORN	53155-8	µmol/L
Asparagine + Ornithine / Phenylalanine Ratio	[ASN + ORN] / PHE	53396-8	molar ratio
Asparagine + Ornithine / Serine Ratio	[ASN + ORN] / SER	53395-0	molar ratio

Citrulline / Phenylalanine Ratio	CIT / PHE	53157-4	molar ratio
Citrulline / Tyrosine Ratio	CIT / TYR	53399-2	molar ratio
Methionine / Alloisoleucine + Isoleucine + Leucine + Hydroxyproline Ratio	MET / [AILE + ILE + LEU + OHPRO]	53397-6	molar ratio
Methionine / Phenylalanine Ratio	MET / PHE	53156-6	molar ratio
Oxoproline + Pipecolate	OXOPRO + PIPA	53232-5	µmol/L
Oxoproline + Pipecolate / Phenylalanine Ratio	[OXOPRO + PIPA] / PHE	53394-3	molar ratio
Phenylalanine / Tyrosine Ratio	PHE / TYR	35572-7	molar ratio
Proline / Phenylalanine Ratio	PRO / PHE	53392-7	TBD
Valine / Phenylalanine Ratio	VAL/PHE	53151-7	molar ratio
Valine + Alloisoleucine + Isoleucine + Leucine + Hydroxyproline + Valine / Phenylalanine + Tyrosine Ratio	[AILE + ILE + LEU + OHPRO + VAL] /	53393-5	molar ratio

MS/MS: Acyl-Carnitine

Analyte	Short Name	LOINC Code	Units
Decatrienoylcarnitine	C10:3	53208-5	µmol/L
Dehydrosebacylcarnitine	C10:1DC	53211-9	µmol/L
Dehydrosuberylcarnitine	C8:1DC	53209-3	µmol/L
Dicarboxydodecanoylcarnitine	C12DC	53214-3	µmol/L
Dicarboxydodecenoylcarnitine	C12:1DC	53213-5	µmol/L
Dicarboxyoleylcarnitine	C18:1DC	53219-2	µmol/L
Dicarboxypalmitoleylcarnitine	C16:1DC	53217-6	µmol/L
Dicarboxypalmitoylcarnitine	C16DC	53218-4	µmol/L
Dicarboxystearoylcarnitine	C18DC	53220-0	µmol/L
Dicarboxytetradecanoylcarnitine	C14DC	53216-8	µmol/L
Dicarboxytetradecenoylcarnitine	C14:1DC	53215-0	µmol/L

Heptanoylcarnitine	C7	53204-4	µmol/L
Hexenoylcarnitine	C6:1	53203-6	µmol/L
Nonanoylcarnitine	C9	53207-7	µmol/L
Octenoylcarnitine	C8:1	53174-9	µmol/L
Phenylacetylcarnitine	PHEC2	53205-1	µmol/L
Salicylylcarnitine	SALC	53206-9	µmol/L
Sebacylcarnitine	C10DC	53212-7	µmol/L
Suberylcarnitine	C8DC	53210-1	µmol/L

MS/MS: Fatty Acid Oxidase

Analyte	Short Name	LOINC Code	Units
Carnitine.free	C0	38481-8	µmol/L
Decadienoylcarnitine	C10:2	53180-6	µmol/L
Decanoylcarnitine	C10	45197-1	µmol/L
Decenoylcarnitine	C10:1	45198-9	µmol/L
Dodecanoylcarnitine	C12	45199-7	µmol/L
Dodecenoylcarnitine	C12:1	45200-3	µmol/L
Hexanoylcarnitine	C6	45211-0	µmol/L
Hydroxybutyrylcarnitine	C4OH	50102-3	µmol/L
Hydroxydecenoylcarnitine	C10:1OH	53182-2	µmol/L
Hydroxydodecanoylcarnitine	C12OH	53189-7	µmol/L
Hydroxydodecenoylcarnitine	C12:1OH	53188-9	µmol/L
Hydroxyhexanoylcarnitine	C6OH	53173-1	µmol/L
Hydroxylinoleoylcarnitine	C18:2OH	50109-8	µmol/L

Hydroxyoleylcarnitine	C18:1OH	50113-0	µmol/L
Hydroxypalmitoleylcarnitine	C16:1OH	50121-3	µmol/L
Hydroxypalmitoylcarnitine	C16OH	50125-4	µmol/L
Hydroxystearoylcarnitine	C18OH	50132-0	µmol/L
Hydroxytetradecadienylcarnitine	C14:2OH	53196-2	µmol/L
Hydroxytetradecanoylcarnitine	C14OH	50281-5	µmol/L
Hydroxytetradecenoylcarnitine	C14:1OH	53197-0	µmol/L
Linoleoylcarnitine	C18:2	45217-7	µmol/L
Octanoylcarnitine	C8	53175-6	µmol/L
Oleylcarnitine	C18:1	53202-8	µmol/L
Palmitoleylcarnitine	C16:1	53198-8	µmol/L
Palmitoylcarnitine	C16	53199-6	µmol/L
Stearoylcarnitine	C18	53241-6	µmol/L
Tetradecadienoylcarnitine	C14:2	53190-5	µmol/L
Tetradecanoylcarnitine	C14	53192-1	µmol/L
Tetradecenoylcarnitine	C14:1	53191-3	µmol/L

MS/MS: Fatty Acid Oxidase Computed Sums and Ratios

Analyte	Short Name	LOINC Code	Units
Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio	C0 / [C16 + C18]	53235-8	molar ratio
Carnitine.free / Palmitoylcarnitine Ratio	C0 / C16	53233-3	molar ratio
Carnitine.free / Stearoylcarnitine Ratio	C0 / C18	53234-1	molar ratio
Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio	[C0 + C2 + C3 + C16 + C18:1 + C18]	53236-6	molar ratio
Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio	C16OH / C16	53201-0	molar ratio

Octanoylcarnitine / Acetylcarnitine Ratio	C8 / C2	53176-4	molar ratio
Octanoylcarnitine / Decanoylcarnitine Ratio	C8 / C10	53177-2	molar ratio
Stearoylcarnitine / Propionylcarnitine Ratio	C18 / C3	53400-8	molar ratio
Tetradecenoylcarnitine / Palmitoylcarnitine Ratio	C14:1 / C16	53195-4	molar ratio
Tetradecenoylcarnitine / Acetylcarnitine Ratio	C14:1 / C2	53193-9	molar ratio
Tetradecenoylcarnitine / Dodecenoylcarnitine Ratio	C14:1 / C12:1	53194-7	molar ratio

MS/MS: Fatty Acid Oxidase-Organic Acids

Analyte	Short Name	LOINC Code	Units
Acetylcarnitine	C2	50157-7	µmol/L

MS/MS: Fatty Acid Oxidase-Organic Acids Computed Sums and Ratios

Analyte	Short Name	LOINC Code	Units
Butyrylcarnitine + Isobutyrylcarnitine	C4	53166-5	µmol/L
Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio	C4 / C2	53167-3	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio	C4 / C8	53169-9	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio	C4 / C3	53168-1	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine	C5DC + C10OH	53183-0	µmol/L
Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio	C5DC + C10OH / C5OH	53184-8	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio	C5DC + C10OH / C8	53185-5	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio	C5DC + C10OH / C16	53186-3	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine /Butyrylcarnitine + Isobutyrylcarnitine Ratio	C5DC + C10OH / C4	53403-2	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine	C8OH + C3DC	53178-0	µmol/L
Hydroxyoctanoylcarnitine + Malonylcarnitine / Butyrylcarnitine + Isobutyrylcarnitine Ratio	C8OH + C3DC / C4	53402-4	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio	C8OH + C3DC / C10	53179-8	molar ratio

MS/MS: Organic Acids

Analyte	Short Name	LOINC Code	Units
Formiminoglutamic acid	FIGLU	53165-7	µmol/L
Hydroxyisovalerylcarnitine	C5OH	50106-4	µmol/L
Methylglutarylcarnitine	C6DC	53187-1	µmol/L
Methylmalonylcarnitine	C4DC	45222-7	µmol/L
Propenoylcarnitine	C3:1	53237-4	µmol/L
Propionylcarnitine	C3	53160-8	µmol/L
Tiglylcarnitine	C5:1	53170-7	µmol/L

MS/MS: Organic Acids Computed Sums and Ratios

Analyte	Short Name	LOINC Code	Units
Hydroxyisovalerylcarnitine / Carnitine.free Ratio	C5OH / C0	53171-5	molar ratio
Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio	C50H / C8	53172-3	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine	C5	45216-9	µmol/L
Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio	C5 / C2	53239-0	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio	C5 / C0	53238-2	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Octanoylcarnitine Ratio	C5 / C8	53401-6	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio	C5 / C3	53240-8	molar ratio
Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio	C4DC / C5OH	53181-4	molar ratio
Propionylcarnitine / Acetylcarnitine Ratio	C3 / C2	53163-2	molar ratio
Propionylcarnitine / Carnitine.free Ratio	C3 / C0	53162-4	molar ratio
Propionylcarnitine / Methionine Ratio	C3 / MET	53161-6	molar ratio
Propionylcarnitine / Palmitoylcarnitine Ratio	C3 / C16	53164-0	molar ratio

Non-MS/MS:

Analyte	Short Name	LOINC Code	Units
Biotinidase	BIO	TBD	TBD

Non-MS/MS: Cystic Fibrosis

Analyte	Short Name	LOINC Code	Units
CFTR mutation variant panel	TBD	TBD	Specific alleles
DNA sequencing of the CFTR gene	TBD	TBD	TBD
Immunoreactive trypsinogen	IRT	TBD	ng/mL
Sweat chloride	TBD	TBD	TBD

Non-MS/MS: Endocrine Disorders

Analyte	Short Name	LOINC Code	Units
11-deoxycortisol	TBD	53338-0	ng/mL
17-hydroxy progesterone	17OHP	38473-5	ng/mL
21-deoxycortisol	TBD	53341-4	ng/mL
Androstenedione	TBD	53343-0	ng/mL
Cortisol	TBD	53345-5	ng/mL
Deoxycorticosterone	TBD	53347-1	ng/mL
T4	T4	53349-7	ng/dL
TSH	TSH	29575-8	mIU/L

Non-MS/MS: Endocrine Disorders Computed Sums and Ratios

Analyte	Short Name	LOINC Code	Units
(17OHP+Androstenedione)/cortisol	TBD	53336-4	TBD

Non-MS/MS: Galactosemia

Analyte	Short Name	LOINC Code	Units
Enzyme NADPH5	TBD	TBD	mg/dL
Galactose	TBD	TBD	mg/dL

Non-MS/MS: Hemoglobin Disorders

Analyte	Short Name	LOINC Code	Units
Electrophoresis (Cellulose Acetate and Citrate Agar)	Electrophoresis	TBD	Disorder
High Performance Liquid Chromatography	HPLC	TBD	Disorder
Isoelectric Focusing	IEF	TBD	Disorder
Percent Hemoglobin A	%HgB A	TBD	%
Percent Hemoglobin B	%HgB B	TBD	%
Percent Hemoglobin Barts	%HgB Barts	TBD	%
Percent Hemoglobin C	%HgB C	TBD	%
Percent Hemoglobin D	%HgB D	TBD	%
Percent Hemoglobin E	%HgB E	TBD	%
Percent Hemoglobin F	%HgB F	TBD	%
Percent Hemoglobin OARAB	%HgB OARAB	TBD	%
Percent Hemoglobin S	%HgB S	TBD	%

Non-MS/MS: Infectious Diseases

Analyte	Short Name	LOINC Code	Units
Human immunodeficiency virus IgG antibodies	TBD	TBD	Pos or Neg
Toxoplasmosis IgG	TBD	TBD	Pos or Neg
Toxoplasmosis IgM antibodies	TBD	TBD	Pos or Neg

MS/MS Analytes

Ordered By Molecular Weight.

Analyte Categories :
AA - Amino Acids
AC - Acyl-Carnitine
FAO - Fatty Acid Oxidase
FAO-OA - Fatty Acid Oxidase - Organic Acids
OA - Organic Acids

Analyte	Category	Short Name	LOINC Code	Units
Glycine	AA	GLY	47633-3	µmol/L
Alanine + Beta Alanine + Sarcosine	AA	ALA + BALA + SARC	53150-9	µmol/L
Succinylacetone	AA	SUAC	53231-7	µmol/L
Serine	AA	SER	47742-2	µmol/L
Proline	AA	PRO	47732-3	µmol/L
Proline / Phenylalanine Ratio	AA	PRO / PHE	53392-7	TBD
Valine	AA	VAL	47799-2	µmol/L
Valine / Phenylalanine Ratio	AA	VAL/PHE	53151-7	molar ratio
Valine + Alloisoleucine + Isoleucine + Leucine + Hydroxyproline + Valine / Phenylalanine + Tyrosine Ratio	AA	[AILE + ILE + LEU + OHPRO + VAL] /	53393-5	molar ratio
Threonine	AA	THR	47784-4	µmol/L
Oxoproline + Pipecolate	AA	OXOPRO + PIPA	53232-5	µmol/L
Oxoproline + Pipecolate / Phenylalanine Ratio	AA	[OXOPRO + PIPA] / PHE	53394-3	molar ratio
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline	AA	AILE + ILE + LEU + OHPRO	53152-5	µmol/L
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio	AA	[AILE + ILE + LEU + OHPRO] / PHE	53153-3	molar ratio
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine	AA	[AILE + ILE + LEU + OHPRO] / ALA	53154-1	molar ratio
Asparagine + Ornithine	AA	ASN + ORN	53155-8	µmol/L
Asparagine + Ornithine / Serine Ratio	AA	[ASN + ORN] / SER	53395-0	molar ratio
Asparagine + Ornithine / Phenylalanine Ratio	AA	[ASN + ORN] / PHE	53396-8	molar ratio
Lysine	AA	LYS	47689-5	µmol/L

Analyte	Category	Short Name	LOINC Code	Units
Methionine	AA	MET	47700-0	µmol/L
Methionine / Phenylalanine Ratio	AA	MET / PHE	53156-6	molar ratio
Methionine / Alloisoleucine + Isoleucine + Leucine + Hydroxyproline Ratio	AA	MET / [AILE + ILE + LEU + OHPRO]	53397-6	molar ratio
Histidine	AA	HIS	47643-2	µmol/L
Carnitine.free	FAO	C0	38481-8	µmol/L
Carnitine.free / Palmitoylcarnitine Ratio	FAO	C0 / C16	53233-3	molar ratio
Carnitine.free / Stearoylcarnitine Ratio	FAO	C0 / C18	53234-1	molar ratio
Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio	FAO	C0 / [C16 + C18]	53235-8	molar ratio
Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio	FAO	[C0 + C2 + C3 + C16 + C18:1 + C18]	53236-6	molar ratio
Phenylalanine	AA	PHE	29573-3	µmol/L
Phenylalanine / Tyrosine Ratio	AA	PHE / TYR	35572-7	molar ratio
Arginine	AA	ARG	47562-4	µmol/L
Arginine / Phenylalanine Ratio	AA	ARG / PHE	53398-4	TBD
Citrulline	AA	CIT	42892-0	µmol/L
Citrulline / Phenylalanine Ratio	AA	CIT / PHE	53157-4	molar ratio
Citrulline / Tyrosine Ratio	AA	CIT / TYR	53399-2	molar ratio
Tyrosine	AA	TYR	35571-9	µmol/L
Aspartate	AA	ASP	47573-1	µmol/L
Homocitrulline	AA	HOMOCIT	53158-2	µmol/L
Acetylcarnitine	FAO-OA	C2	50157-7	µmol/L
Glutamate	AA	GLU	47623-4	µmol/L
Tryptophan	AA	TRP	53159-0	µmol/L
Propenoylcarnitine	OA	C3:1	53237-4	µmol/L

Analyte	Category	Short Name	LOINC Code	Units
Propionylcarnitine	OA	C3	53160-8	µmol/L
Propionylcarnitine / Methionine Ratio	OA	C3 / MET	53161-6	molar ratio
Propionylcarnitine / Carnitine.free Ratio	OA	C3 / C0	53162-4	molar ratio
Propionylcarnitine / Acetylcarnitine Ratio	OA	C3 / C2	53163-2	molar ratio
Propionylcarnitine / Palmitoylcarnitine Ratio	OA	C3 / C16	53164-0	molar ratio
Formiminoglutamic acid	OA	FIGLU	53165-7	µmol/L
Butyrylcarnitine + Isobutyrylcarnitine	FAO-OA	C4	53166-5	µmol/L
Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio	FAO-OA	C4 / C2	53167-3	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio	FAO-OA	C4 / C3	53168-1	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio	FAO-OA	C4 / C8	53169-9	molar ratio
Tiglylcarnitine	OA	C5:1	53170-7	µmol/L
Isovalerylcarnitine + Methylbutyrylcarnitine	OA	C5	45216-9	µmol/L
Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio	OA	C5 / C0	53238-2	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio	OA	C5 / C2	53239-0	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio	OA	C5 / C3	53240-8	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Octanoylcarnitine Ratio	OA	C5 / C8	53401-6	molar ratio
Hydroxybutyrylcarnitine	FAO	C4OH	50102-3	µmol/L
Hexanoylcarnitine	FAO	C6	45211-0	µmol/L
Hydroxyisovalerylcarnitine	OA	C5OH	50106-4	µmol/L
Hydroxyisovalerylcarnitine / Carnitine.free Ratio	OA	C5OH / C0	53171-5	molar ratio
Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio	OA	C50H / C8	53172-3	molar ratio
Hydroxyhexanoylcarnitine	FAO	C6OH	53173-1	µmol/L
Octenoylcarnitine	AC	C8:1	53174-9	µmol/L

Analyte	Category	Short Name	LOINC Code	Units
Octanoylcarnitine	FAO	C8	53175-6	µmol/L
Octanoylcarnitine / Acetylcarnitine Ratio	FAO	C8 / C2	53176-4	molar ratio
Octanoylcarnitine / Decanoylcarnitine Ratio	FAO	C8 / C10	53177-2	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine	FAO-OA	C8OH + C3DC	53178-0	µmol/L
Hydroxyoctanoylcarnitine + Malonylcarnitine / Butyrylcarnitine + Isobutyrylcarnitine Ratio	FAO-OA	C8OH + C3DC / C4	53402-4	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio	FAO-OA	C8OH + C3DC / C10	53179-8	molar ratio
Decadienoylcarnitine	FAO	C10:2	53180-6	µmol/L
Decenoylcarnitine	FAO	C10:1	45198-9	µmol/L
Decanoylcarnitine	FAO	C10	45197-1	µmol/L
Methylmalonylcarnitine	OA	C4DC	45222-7	µmol/L
Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio	OA	C4DC / C5OH	53181-4	molar ratio
Hydroxydecenoylcarnitine	FAO	C10:1OH	53182-2	µmol/L
Glutarylcarnitine + Hydroxydecanoylcarnitine	FAO-OA	C5DC + C10OH	53183-0	µmol/L
Glutarylcarnitine + Hydroxydecanoylcarnitine /Butyrylcarnitine + Isobutyrylcarnitine Ratio	FAO-OA	C5DC + C10OH / C4	53403-2	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio	FAO-OA	C5DC + C10OH / C5OH	53184-8	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio	FAO-OA	C5DC + C10OH / C8	53185-5	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio	FAO-OA	C5DC + C10OH / C16	53186-3	molar ratio
Dodecenoylcarnitine	FAO	C12:1	45200-3	µmol/L
Dodecanoylcarnitine	FAO	C12	45199-7	µmol/L
Methylglutarylcarnitine	OA	C6DC	53187-1	µmol/L
Hydroxydodecenoylcarnitine	FAO	C12:1OH	53188-9	µmol/L
Hydroxydodecanoylcarnitine	FAO	C12OH	53189-7	µmol/L
Tetradecadienoylcarnitine	FAO	C14:2	53190-5	µmol/L

Analyte	Category	Short Name	LOINC Code	Units
Tetradecenoylcarnitine	FAO	C14:1	53191-3	µmol/L
Tetradecanoylcarnitine	FAO	C14	53192-1	µmol/L
Tetradecenoylcarnitine / Acetylcarnitine Ratio	FAO	C14:1 / C2	53193-9	molar ratio
Tetradecenoylcarnitine / Dodecenoylcarnitine Ratio	FAO	C14:1 / C12:1	53194-7	molar ratio
Tetradecenoylcarnitine / Palmitoylcarnitine Ratio	FAO	C14:1 / C16	53195-4	molar ratio
Hydroxytetradecadienylcarnitine	FAO	C14:2OH	53196-2	µmol/L
Hydroxytetradecenoylcarnitine	FAO	C14:1OH	53197-0	µmol/L
Hydroxytetradecanoylcarnitine	FAO	C14OH	50281-5	µmol/L
Palmitoleylcarnitine	FAO	C16:1	53198-8	µmol/L
Palmitoylcarnitine	FAO	C16	53199-6	µmol/L
Argininosuccinate	AA	ASA	53062-6	µmol/L
Argininosuccinate / Arginine Ratio	AA	ASA / ARG	53200-2	molar ratio
Hydroxypalmitoleylcarnitine	FAO	C16:1OH	50121-3	µmol/L
Hydroxypalmitoylcarnitine	FAO	C16OH	50125-4	µmol/L
Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio	FAO	C16OH / C16	53201-0	molar ratio
Linoleoylcarnitine	FAO	C18:2	45217-7	µmol/L
Oleylcarnitine	FAO	C18:1	53202-8	µmol/L
Stearoylcarnitine	FAO	C18	53241-6	µmol/L
Stearoylcarnitine / Propionylcarnitine Ratio	FAO	C18 / C3	53400-8	molar ratio
Hydroxylinoleoylcarnitine	FAO	C18:2OH	50109-8	µmol/L
Hydroxyoleylcarnitine	FAO	C18:1OH	50113-0	µmol/L
Hydroxystearoylcarnitine	FAO	C18OH	50132-0	µmol/L
Methylhistidine	AA	CH3HIS	47539-2	µmol/L

Analyte	Category	Short Name	LOINC Code	Units
Hexenoylcarnitine	AC	C6:1	53203-6	µmol/L
Heptanoylcarnitine	AC	C7	53204-4	µmol/L
Phenylacetylcarnitine	AC	PHEC2	53205-1	µmol/L
Salicylylcarnitine	AC	SALC	53206-9	µmol/L
Nonanoylcarnitine	AC	C9	53207-7	µmol/L
Decatrienoylcarnitine	AC	C10:3	53208-5	µmol/L
Dehydrosuberylcarnitine	AC	C8:1DC	53209-3	µmol/L
Suberylcarnitine	AC	C8DC	53210-1	µmol/L
Dehydrosebacylcarnitine	AC	C10:1DC	53211-9	µmol/L
Sebacylcarnitine	AC	C10DC	53212-7	µmol/L
Dicarboxydodecenoylcarnitine	AC	C12:1DC	53213-5	µmol/L
Dicarboxydodecanoylcarnitine	AC	C12DC	53214-3	µmol/L
Dicarboxytetradecenoylcarnitine	AC	C14:1DC	53215-0	µmol/L
Dicarboxytetradecanoylcarnitine	AC	C14DC	53216-8	µmol/L
Dicarboxypalmitoleylcarnitine	AC	C16:1DC	53217-6	µmol/L
Dicarboxypalmitoylcarnitine	AC	C16DC	53218-4	µmol/L
Dicarboxyoleylcarnitine	AC	C18:1DC	53219-2	µmol/L
Dicarboxystearoylcarnitine	AC	C18DC	53220-0	µmol/L

MS/MS Analytes

Grouped by Computed Sum or Ratio.
Ordered by Molecular Weight.

Analyte Categories :
- AA - Amino Acids
- AC - Acyl-Carnitine
- FAO - Fatty Acid Oxidase
- FAO-OA - Fatty Acid Oxidase - Organic Acids
- OA - Organic Acids

Analyte	Category	Short Name	LOINC Code	Units
Glycine	AA	GLY	47633-3	µmol/L
Succinylacetone	AA	SUAC	53231-7	µmol/L
Serine	AA	SER	47742-2	µmol/L
Proline	AA	PRO	47732-3	µmol/L
Valine	AA	VAL	47799-2	µmol/L
Threonine	AA	THR	47784-4	µmol/L
Lysine	AA	LYS	47689-5	µmol/L
Methionine	AA	MET	47700-0	µmol/L
Histidine	AA	HIS	47643-2	µmol/L
Carnitine.free	FAO	C0	38481-8	µmol/L
Phenylalanine	AA	PHE	29573-3	µmol/L
Arginine	AA	ARG	47562-4	µmol/L
Citrulline	AA	CIT	42892-0	µmol/L
Tyrosine	AA	TYR	35571-9	µmol/L
Aspartate	AA	ASP	47573-1	µmol/L
Homocitrulline	AA	HOMOCIT	53158-2	µmol/L
Acetylcarnitine	FAO-OA	C2	50157-7	µmol/L

Glutamate	AA	GLU	47623-4	µmol/L
Tryptophan	AA	TRP	53159-0	µmol/L
Propenoylcarnitine	OA	C3:1	53237-4	µmol/L
Propionylcarnitine	OA	C3	53160-8	µmol/L
Formiminoglutamic acid	OA	FIGLU	53165-7	µmol/L
Tiglylcarnitine	OA	C5:1	53170-7	µmol/L
Hydroxybutyrylcarnitine	FAO	C4OH	50102-3	µmol/L
Hexanoylcarnitine	FAO	C6	45211-0	µmol/L
Hydroxyisovalerylcarnitine	OA	C5OH	50106-4	µmol/L
Hydroxyhexanoylcarnitine	FAO	C6OH	53173-1	µmol/L
Octenoylcarnitine	AC	C8:1	53174-9	µmol/L
Octanoylcarnitine	FAO	C8	53175-6	µmol/L
Decadienoylcarnitine	FAO	C10:2	53180-6	µmol/L
Decenoylcarnitine	FAO	C10:1	45198-9	µmol/L
Decanoylcarnitine	FAO	C10	45197-1	µmol/L
Methylmalonylcarnitine	OA	C4DC	45222-7	µmol/L
Hydroxydecenoylcarnitine	FAO	C10:1OH	53182-2	µmol/L
Dodecenoylcarnitine	FAO	C12:1	45200-3	µmol/L
Dodecanoylcarnitine	FAO	C12	45199-7	µmol/L
Methylglutarylcarnitine	OA	C6DC	53187-1	µmol/L
Hydroxydodecenoylcarnitine	FAO	C12:1OH	53188-9	µmol/L
Hydroxydodecanoylcarnitine	FAO	C12OH	53189-7	µmol/L
Tetradecadienoylcarnitine	FAO	C14:2	53190-5	µmol/L
Tetradecenoylcarnitine	FAO	C14:1	53191-3	µmol/L

Tetradecanoylcarnitine	FAO	C14	53192-1	µmol/L
Hydroxytetradecadienylcarnitine	FAO	C14:2OH	53196-2	µmol/L
Hydroxytetradecenoylcarnitine	FAO	C14:1OH	53197-0	µmol/L
Hydroxytetradecanoylcarnitine	FAO	C14OH	50281-5	µmol/L
Palmitoleylcarnitine	FAO	C16:1	53198-8	µmol/L
Palmitoylcarnitine	FAO	C16	53199-6	µmol/L
Argininosuccinate	AA	ASA	53062-6	µmol/L
Hydroxypalmitoleylcarnitine	FAO	C16:1OH	50121-3	µmol/L
Hydroxypalmitoylcarnitine	FAO	C16OH	50125-4	µmol/L
Linoleoylcarnitine	FAO	C18:2	45217-7	µmol/L
Oleylcarnitine	FAO	C18:1	53202-8	µmol/L
Stearoylcarnitine	FAO	C18	53241-6	µmol/L
Hydroxylinoleoylcarnitine	FAO	C18:2OH	50109-8	µmol/L
Hydroxyoleylcarnitine	FAO	C18:1OH	50113-0	µmol/L
Hydroxystearoylcarnitine	FAO	C18OH	50132-0	µmol/L
Methylhistidine	AA	CH3HIS	47539-2	µmol/L
Hexenoylcarnitine	AC	C6:1	53203-6	µmol/L
Heptanoylcarnitine	AC	C7	53204-4	µmol/L
Phenylacetylcarnitine	AC	PHEC2	53205-1	µmol/L
Salicylylcarnitine	AC	SALC	53206-9	µmol/L
Nonanoylcarnitine	AC	C9	53207-7	µmol/L
Decatrienoylcarnitine	AC	C10:3	53208-5	µmol/L
Dehydrosuberylcarnitine	AC	C8:1DC	53209-3	µmol/L
Suberylcarnitine	AC	C8DC	53210-1	µmol/L

Dehydrosebacylcarnitine	AC	C10:1DC	53211-9	µmol/L
Sebacylcarnitine	AC	C10DC	53212-7	µmol/L
Dicarboxydodecenoylcarnitine	AC	C12:1DC	53213-5	µmol/L
Dicarboxydodecanoylcarnitine	AC	C12DC	53214-3	µmol/L
Dicarboxytetradecenoylcarnitine	AC	C14:1DC	53215-0	µmol/L
Dicarboxytetradecanoylcarnitine	AC	C14DC	53216-8	µmol/L
Dicarboxypalmitoleylcarnitine	AC	C16:1DC	53217-6	µmol/L
Dicarboxypalmitoylcarnitine	AC	C16DC	53218-4	µmol/L
Dicarboxyoleylcarnitine	AC	C18:1DC	53219-2	µmol/L
Dicarboxystearoylcarnitine	AC	C18DC	53220-0	µmol/L

Computed Sums and Ratios

Analyte	Category	Short Name	LOINC Code	Units
Alanine + Beta Alanine + Sarcosine	AA	ALA + BALA + SARC	53150-9	µmol/L
Proline / Phenylalanine Ratio	AA	PRO / PHE	53392-7	TBD
Valine / Phenylalanine Ratio	AA	VAL/PHE	53151-7	molar ratio
Valine + Alloisoleucine + Isoleucine + Leucine + Hydroxyproline + Valine / Phenylalanine + Tyrosine Ratio	AA	[AILE + ILE + LEU + OHPRO + VAL] /	53393-5	molar ratio
Oxoproline + Pipecolate	AA	OXOPRO + PIPA	53232-5	µmol/L
Oxoproline + Pipecolate / Phenylalanine Ratio	AA	[OXOPRO + PIPA] / PHE	53394-3	molar ratio
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline	AA	AILE + ILE + LEU + OHPRO	53152-5	µmol/L
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio	AA	[AILE + ILE + LEU + OHPRO] / PHE	53153-3	molar ratio
Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine	AA	[AILE + ILE + LEU + OHPRO] / ALA	53154-1	molar ratio
Asparagine + Ornithine	AA	ASN + ORN	53155-8	µmol/L
Asparagine + Ornithine / Serine Ratio	AA	[ASN + ORN] / SER	53395-0	molar ratio

Asparagine + Ornithine / Phenylalanine Ratio	AA	[ASN + ORN] / PHE	53396-8	molar ratio
Methionine / Phenylalanine Ratio	AA	MET / PHE	53156-6	molar ratio
Methionine / Alloisoleucine + Isoleucine + Leucine + Hydroxyproline Ratio	AA	MET / [AILE + ILE + LEU + OHPRO]	53397-6	molar ratio
Carnitine.free / Palmitoylcarnitine Ratio	FAO	C0 / C16	53233-3	molar ratio
Carnitine.free / Stearoylcarnitine Ratio	FAO	C0 / C18	53234-1	molar ratio
Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio	FAO	C0 / [C16 + C18]	53235-8	molar ratio
Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio	FAO	[C0 + C2 + C3 + C16 + C18:1 + C18]	53236-6	molar ratio
Phenylalanine / Tyrosine Ratio	AA	PHE / TYR	35572-7	molar ratio
Arginine / Phenylalanine Ratio	AA	ARG / PHE	53398-4	TBD
Citrulline / Phenylalanine Ratio	AA	CIT / PHE	53157-4	molar ratio
Citrulline / Tyrosine Ratio	AA	CIT / TYR	53399-2	molar ratio
Propionylcarnitine / Methionine Ratio	OA	C3 / MET	53161-6	molar ratio
Propionylcarnitine / Carnitine.free Ratio	OA	C3 / C0	53162-4	molar ratio
Propionylcarnitine / Acetylcarnitine Ratio	OA	C3 / C2	53163-2	molar ratio
Propionylcarnitine / Palmitoylcarnitine Ratio	OA	C3 / C16	53164-0	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine	FAO-OA	C4	53166-5	µmol/L
Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio	FAO-OA	C4 / C2	53167-3	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio	FAO-OA	C4 / C3	53168-1	molar ratio
Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio	FAO-OA	C4 / C8	53169-9	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine	OA	C5	45216-9	µmol/L
Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio	OA	C5 / C0	53238-2	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio	OA	C5 / C2	53239-0	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio	OA	C5 / C3	53240-8	molar ratio
Isovalerylcarnitine + Methylbutyrylcarnitine / Octanoylcarnitine Ratio	OA	C5 / C8	53401-6	molar ratio

Hydroxyisovalerylcarnitine / Carnitine.free Ratio	OA	C5OH / C0	53171-5	molar ratio
Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio	OA	C50H / C8	53172-3	molar ratio
Octanoylcarnitine / Acetylcarnitine Ratio	FAO	C8 / C2	53176-4	molar ratio
Octanoylcarnitine / Decanoylcarnitine Ratio	FAO	C8 / C10	53177-2	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine	FAO-OA	C8OH + C3DC	53178-0	µmol/L
Hydroxyoctanoylcarnitine + Malonylcarnitine / Butyrylcarnitine + Isobutyrylcarnitine Ratio	FAO-OA	C8OH + C3DC / C4	53402-4	molar ratio
Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio	FAO-OA	C8OH + C3DC / C10	53179-8	molar ratio
Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio	OA	C4DC / C5OH	53181-4	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine	FAO-OA	C5DC + C10OH	53183-0	µmol/L
Glutarylcarnitine + Hydroxydecanoylcarnitine /Butyrylcarnitine + Isobutyrylcarnitine Ratio	FAO-OA	C5DC + C10OH / C4	53403-2	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio	FAO-OA	C5DC + C10OH / C5OH	53184-8	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio	FAO-OA	C5DC + C10OH / C8	53185-5	molar ratio
Glutarylcarnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio	FAO-OA	C5DC + C10OH / C16	53186-3	molar ratio
Tetradecenoylcarnitine / Acetylcarnitine Ratio	FAO	C14:1 / C2	53193-9	molar ratio
Tetradecenoylcarnitine / Dodecenoylcarnitine Ratio	FAO	C14:1 / C12:1	53194-7	molar ratio
Tetradecenoylcarnitine / Palmitoylcarnitine Ratio	FAO	C14:1 / C16	53195-4	molar ratio
Argininosuccinate / Arginine Ratio	AA	ASA / ARG	53200-2	molar ratio
Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio	FAO	C16OH / C16	53201-0	molar ratio
Stearoylcarnitine / Propionylcarnitine Ratio	FAO	C18 / C3	53400-8	molar ratio

MS/MS Condition to Analyte Mapping

The MS/MS Condition to Analyte Mapping report lists the analytes or laboratory measures that may be abnormal in the presence of a condition or disorder (listed in boldface). The less common abnormal findings for each condition are marked as optional. The mapping of abnormal analytes to specific conditions is not precise and this listing is intended to guide looking for specific laboratory abnormalities when a specific condition is suspected.

*** Denotes an Optional Analyte**

MS/MS: ACMG Primary Targets: Amino Acids

Argininosuccinic aciduria (ASA)

Argininosuccinate (ASA)

Argininosuccinate / Arginine Ratio (ASA / ARG)

Citrulline (CIT)

Citrulline / Phenylalanine Ratio (CIT / PHE)

Citrullinemia type I (CIT I)

Citrulline (CIT)

Citrulline / Phenylalanine Ratio (CIT / PHE)

Homocystinuria (HCY)

Methionine (MET)

Methionine / Phenylalanine Ratio (MET / PHE)

Maple syrup urine disease (MSUD)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline (AILE + ILE + LEU + OHPRO)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio ([AILE + ILE + LEU + OHPRO] / PHE)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine ([AILE + ILE + LEU + OHPRO] / ALA)

Valine (VAL)

Valine / Phenylalanine Ratio (VAL/PHE)

Phenylketonuria (PKU)

Phenylalanine (PHE)

Phenylalanine / Tyrosine Ratio (PHE / TYR)

Tyrosinemia type I (TYR I)

Succinylacetone (SUAC)

Tyrosine (TYR)*

MS/MS: ACMG Primary Targets: Fatty Acid Oxidase

Carnitine uptake defect (CUD)

Acetylcarnitine (C2)

Carnitine.free (C0)

Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio ([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)

Linoleoylcarnitine (C18:2)*

Oleylcarnitine (C18:1)*

Palmitoylcarnitine (C16)*

Stearoylcarnitine (C18)*

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Hydroxydecenoylcarnitine (C10:1OH)*

Hydroxydodecanoylcarnitine (C12OH)*

Hydroxydodecenoylcarnitine (C12:1OH)*

Hydroxylinoleoylcarnitine (C18:2OH)

Hydroxyoleylcarnitine (C18:1OH)

Hydroxypalmitoleylcarnitine (C16:1OH)

Hydroxypalmitoylcarnitine (C16OH)

Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio (C16OH / C16)

Hydroxytetradecadienylcarnitine (C14:2OH)*

Hydroxytetradecanoylcarnitine (C14OH)

Hydroxytetradecenoylcarnitine (C14:1OH)*

Linoleoylcarnitine (C18:2)*

Oleylcarnitine (C18:1)*

Palmitoleylcarnitine (C16:1)*

Palmitoylcarnitine (C16)

Stearoylcarnitine (C18)*

Stearoylcarnitine / Propionylcarnitine Ratio (C18 / C3)*

Tetradecenoylcarnitine (C14:1)*

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Decanoylcarnitine (C10)

Decenoylcarnitine (C10:1)

Hexanoylcarnitine (C6)*

Octanoylcarnitine (C8)

Octanoylcarnitine / Acetylcarnitine Ratio (C8 / C2)

Octanoylcarnitine / Decanoylcarnitine Ratio (C8 / C10)*

Trifunctional protein deficiency (TFP)

Hydroxydecenoylcarnitine (C10:1OH)*

Hydroxydodecanoylcarnitine (C12OH)*

Hydroxydodecenoylcarnitine (C12:1OH)*

Hydroxylinoleoylcarnitine (C18:2OH)

Hydroxyoleylcarnitine (C18:1OH)

Hydroxypalmitoleylcarnitine (C16:1OH)

Hydroxypalmitoylcarnitine (C16OH)

Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio (C16OH / C16)

Hydroxytetradecadienylcarnitine (C14:2OH)*

Hydroxytetradecanoylcarnitine (C14OH)

Hydroxytetradecenoylcarnitine (C14:1OH)*

Linoleoylcarnitine (C18:2)*

Oleylcarnitine (C18:1)*

Palmitoleylcarnitine (C16:1)*

Palmitoylcarnitine (C16)

Stearoylcarnitine (C18)*

Stearoylcarnitine / Propionylcarnitine Ratio (C18 / C3)*

Tetradecenoylcarnitine (C14:1)*

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Dodecanoylcarnitine (C12)*

Dodecenoylcarnitine (C12:1)*

Palmitoleylcarnitine (C16:1)*

Palmitoylcarnitine (C16)

Tetradecadienoylcarnitine (C14:2)

Tetradecanoylcarnitine (C14)

Tetradecenoylcarnitine (C14:1)

Tetradecenoylcarnitine / Palmitoylcarnitine Ratio (C14:1 / C16)

Tetradecenoylcarnitine / Acetylcarnitine Ratio (C14:1 / C2)

Tetradecenoylcarnitine / Dodecenoylcarnitine Ratio (C14:1 / C12:1)*

MS/MS: ACMG Primary Targets: Organic Acids

3-Hydroxy-3-methylglutaric aciduria (HMG)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)*

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

Methylglutarylcarnitine (C6DC)

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)*

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

beta-Ketothiolase deficiency (BKT)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)*

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

Tiglylcarnitine (C5:1)

Glutaric acidemia type I (GA I)

Glutarylcarnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)

Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio (C5DC + C10OH / C5OH)

Glutarylcarnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio (C5DC + C10OH / C8)

Glutarylcarnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio (C5DC + C10OH / C16)

Isovaleric acidemia (IVA)

Isovalerylcarnitine + Methylbutyrylcarnitine (C5)

Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio (C5 / C2)

Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio (C5 / C0)

Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio (C5 / C3)

Methylmalonic acidemia (CBL A)

Methylmalonylcarnitine (C4DC)*

Methylmalonylcarnitine (C4DC)*

Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio (C4DC / C5OH)*

Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio (C4DC / C5OH)*

Propionylcarnitine (C3)

Propionylcarnitine (C3)

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)*

Multiple carboxylase deficiency (MCD)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)*

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

Propionylcarnitine (C3)*

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)*

Propionic acidemia (PROP)

Carnitine.free (C0)

Propionylcarnitine (C3)

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)*

MS/MS: ACMG Secondary Conditions: Amino Acids

Argininemia (ARG)

Arginine (ARG)

Citrullinemia type II (CIT II)

Arginine (ARG)*

Citrulline (CIT)

Citrulline / Phenylalanine Ratio (CIT / PHE)

Threonine (THR)*

Disorders of biopterin biosynthesis (BIOPT-BIO)

Phenylalanine (PHE)

Phenylalanine / Tyrosine Ratio (PHE / TYR)

Disorders of biopterin regeneration (BIOPT-REG)

Phenylalanine (PHE)

Phenylalanine / Tyrosine Ratio (PHE / TYR)

Hypermethioninemia (MET)

Methionine (MET)

Methionine / Phenylalanine Ratio (MET / PHE)

Hyperphenylalaninemia (variant, benign) (H-PHE)

Phenylalanine (PHE)

Phenylalanine / Tyrosine Ratio (PHE / TYR)

Tyrosinemia type II (TYR II)

Tyrosine (TYR)

Tyrosinemia type III (TYR III)

Tyrosine (TYR)

MS/MS: ACMG Secondary Conditions: Fatty Acid Oxidase

2,4-Dienoyl-CoA reductase deficiency (DE RED)

Decadienoylcarnitine (C10:2)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine.free (C0)

Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio (C0 / [C16 + C18])

Carnitine.free / Palmitoylcarnitine Ratio (C0 / C16)*

Carnitine.free / Stearoylcarnitine Ratio (C0 / C18)*

Linoleoylcarnitine (C18:2)

Oleylcarnitine (C18:1)

Palmitoylcarnitine (C16)

Stearoylcarnitine (C18)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio (C0 / [C16 + C18])*

Linoleoylcarnitine (C18:2)

Oleylcarnitine (C18:1)

Palmitoleylcarnitine (C16:1)*

Palmitoylcarnitine (C16)

Stearoylcarnitine (C18)

Tetradecanoylcarnitine (C14)*

Carnitine-acylcarnitine translocase deficiency (CACT)

Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio (C0 / [C16 + C18])*

Linoleoylcarnitine (C18:2)

Oleylcarnitine (C18:1)

Palmitoleylcarnitine (C16:1)*

Palmitoylcarnitine (C16)

Stearoylcarnitine (C18)

Tetradecanoylcarnitine (C14)*

Glutaric acidemia type II (GA II)

Butyrylcarnitine + Isobutyrylcarnitine (C4)

Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio (C4 / C2)

Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio (C4 / C3)

Decanoylcarnitine (C10)

Decenoylcarnitine (C10:1)

Glutarylcarnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)

Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio (C5DC + C10OH / C5OH)

Hexanoylcarnitine (C6)*

Isovalerylcarnitine + Methylbutyrylcarnitine (C5)

Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio (C5 / C2)

Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio (C5 / C0)

Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio (C5 / C3)

Octanoylcarnitine (C8)

Octanoylcarnitine / Acetylcarnitine Ratio (C8 / C2)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

Decanoylcarnitine (C10)*

Decenoylcarnitine (C10:1)*

Hydroxydecenoylcarnitine (C10:1OH)*

Hydroxyhexanoylcarnitine (C6OH)

Hydroxyoctanoylcarnitine + Malonylcarnitine (C8OH + C3DC)

Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio (C8OH + C3DC / C10)

Octanoylcarnitine (C8)

Octanoylcarnitine / Acetylcarnitine Ratio (C8 / C2)

Octanoylcarnitine / Decanoylcarnitine Ratio (C8 / C10)*

Octenoylcarnitine (C8:1)*

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Butyrylcarnitine + Isobutyrylcarnitine (C4)

Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio (C4 / C2)

Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio (C4 / C8)

Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio (C4 / C3)

Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (SCHAD)

Acetylcarnitine (C2)*

Hydroxybutyrylcarnitine (C4OH)

Hydroxyhexanoylcarnitine (C6OH)

MS/MS: ACMG Secondary Conditions: Organic Acids

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

Tiglylcarnitine (C5:1)

2-Methylbutyrylglycinuria (2MBG)

Isovalerylcarnitine + Methylbutyrylcarnitine (C5)

Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio (C5 / C2)

Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio (C5 / C0)

Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio (C5 / C3)

3-Methylglutaconic aciduria (3MGA)

Hydroxyisovalerylcarnitine (C5OH)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

Isobutyrylglycinuria (IBD)

Butyrylcarnitine + Isobutyrylcarnitine (C4)

Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio (C4 / C2)

Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio (C4 / C8)

Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio (C4 / C3)

Malonic acidemia (MAL)

Hydroxyoctanoylcarnitine + Malonylcarnitine (C8OH + C3DC)

Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio (C8OH + C3DC / C10)

Methylmalonic aciduria and homocystinuria (CBL C)

Methionine (MET)*

Methionine / Phenylalanine Ratio (MET / PHE)*

Methylmalonylcarnitine (C4DC)*

Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio (C4DC / C5OH)*

Propionylcarnitine (C3)

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Methionine Ratio (C3 / MET)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)*

MS/MS: Other Conditions: Amino Acids

Carbamoyltransferase deficiency (CPS)

Citrulline (CIT)*

Citrulline / Phenylalanine Ratio (CIT / PHE)*

Girate atrophy of the retina (Hyper ORN)

Asparagine + Ornithine (ASN + ORN)

Histidinemia (HIS)

Histidine (HIS)

Homocystinuria-megaloblastic anemia (CBL G)

Methionine (MET)

Methionine / Phenylalanine Ratio (MET / PHE)

Hydroxyprolinemia (OH PRO)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline (AILE + ILE + LEU + OHPRO)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio ([AILE + ILE + LEU + OHPRO] / PHE)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine ([AILE + ILE + LEU + OHPRO] / ALA)

Hyperlysinemia (Hyper LYS)

Lysine (LYS)

Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome (HHH)

Asparagine + Ornithine (ASN + ORN)*

Homocitrulline (HOMOCIT)

Methylcobalamin deficiency (CBL E)

Methionine (MET)

Methionine / Phenylalanine Ratio (MET / PHE)

Methylene tetrahydrofolate reductase deficiency (MTHFR)

Methionine (MET)

Methionine / Phenylalanine Ratio (MET / PHE)

Nonketotic hyperglycinemia (glycine encephalopathy) (NKHG)

Glycine (GLY)

Ornithine transcarbamylase deficiency (OTC)

Citrulline (CIT)*

Citrulline / Phenylalanine Ratio (CIT / PHE)*

Pyroglutamic acidemia (OXO PRO)

Oxoproline + Pipecolate (OXOPRO + PIPA)

Pyruvate carboxylase deficiency (PC)

Alanine + Beta Alanine + Sarcosine (ALA + BALA + SARC)*

Citrulline (CIT)

Citrulline / Phenylalanine Ratio (CIT / PHE)

Lysine (LYS)*

Proline (PRO)*

Valinemia (Hyper VAL)

Valine (VAL)

Valine / Phenylalanine Ratio (VAL/PHE)

MS/MS: Other Conditions: Fatty Acid Oxidase

Maternal carnitine uptake defect (CUD (mat))

Carnitine.free (C0)

Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio ([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)

Homocitrulline (HOMOCIT)*

MS/MS: Other Conditions: Organic Acids

Ethylmalonic encephalopathy (EE)

Butyrylcarnitine + Isobutyrylcarnitine (C4)

Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio (C4 / C2)

Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio (C4 / C3)

Formiminoglutamic acid (FIGLU)

Isovalerylcarnitine + Methylbutyrylcarnitine (C5)

Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio (C5 / C2)

Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio (C5 / C0)

Tiglylcarnitine (C5:1)

Formiminoglutamic acidemia (FIGLU)

Homocitrulline (HOMOCIT)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))

Carnitine.free (C0)*

Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio ([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)*

Hexanoylcarnitine (C6)

Homocitrulline (HOMOCIT)*

Hydroxybutyrylcarnitine (C4OH)

Hydroxyisovalerylcarnitine (C5OH)

Maternal glutaric acidemia type I (GA I (mat))

Carnitine.free (C0)

Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio ([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)

Glutarylcarnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)*

Glutarylcarnitine + Hydroxydecanoylcarnitine /Butyrylcarnitine + Isobutyrylcarnitine Ratio (C5DC + C10OH / C4)

Hydroxydecenoylcarnitine (C10:1OH)*

Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio (C4DC / C5OH)*

Primary lactic acidemia (various types) (LACTIC)

Alanine + Beta Alanine + Sarcosine (ALA + BALA + SARC)*

Proline (PRO)*

Succinyl-CoA ligase deficiency (SUCLA2)

Decanoylcarnitine (C10)

Decenoylcarnitine (C10:1)

Propenoylcarnitine (C3:1)

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)*

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)*

Propionylcarnitine / Methionine Ratio (C3 / MET)*

MS/MS Analytes to Conditions Mapping

The MS/MS Analyte to Condition Mapping report lists the conditions or disorders that are suggested by an abnormal analyte or laboratory measurements (listed in boldface). Final diagnosis depends on confirmatory testing and less common conditions are marked as optional. The mapping of abnormal analytes to specific conditions is not precise and this listing is intended to guide further investigation of the cause of the laboratory abnormality.

*** Denotes an Optional Condition**

MS/MS: Amino Acids

Arginine (ARG)

Argininemia (ARG)

Citrullinemia type II (CIT II)*

Argininosuccinate (ASA)

Argininosuccinic aciduria (ASA)

Citrulline (CIT)

Argininosuccinic aciduria (ASA)

Carbamoyltransferase deficiency (CPS)*

Citrullinemia type I (CIT I)

Citrullinemia type II (CIT II)

Ornithine transcarbamylase deficiency (OTC)*

Pyruvate carboxylase deficiency (PC)

Glycine (GLY)

Nonketotic hyperglycinemia (glycine encephalopathy) (NKHG)

Histidine (HIS)

Histidinemia (HIS)

Homocitrulline (HOMOCIT)

Formiminoglutamic acidemia (FIGLU)*

Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome (HHH)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))*

Maternal carnitine uptake defect (CUD (mat))*

Lysine (LYS)

Hyperlysinemia (Hyper LYS)

Pyruvate carboxylase deficiency (PC)*

Methionine (MET)

Homocystinuria (HCY)

Homocystinuria-megaloblastic anemia (CBL G)

Hypermethioninemia (MET)

Methylcobalamin deficiency (CBL E)

Methylene tetrahydrofolate reductase deficiency (MTHFR)

Methylmalonic aciduria and homocystinuria (CBL C)*

Phenylalanine (PHE)

Disorders of biopterin biosynthesis (BIOPT-BIO)

Disorders of biopterin regeneration (BIOPT-REG)

Hyperphenylalaninemia (variant, benign) (H-PHE)

Phenylketonuria (PKU)

Proline (PRO)

Primary lactic acidemia (various types) (LACTIC)*

Pyruvate carboxylase deficiency (PC)*

Succinylacetone (SUAC)

Tyrosinemia type I (TYR I)

Threonine (THR)

Citrullinemia type II (CIT II)*

Tyrosine (TYR)

Tyrosinemia type I (TYR I)*

Tyrosinemia type II (TYR II)

Tyrosinemia type III (TYR III)

Valine (VAL)

Maple syrup urine disease (MSUD)

Valinemia (Hyper VAL)

MS/MS: Amino Acids CALCULATED RATIO

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline /Alanine ([AILE + ILE + LEU + OHPRO] / ALA)

Hydroxyprolinemia (OH PRO)

Maple syrup urine disease (MSUD)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline / Phenylalanine Ratio ([AILE + ILE + LEU + OHPRO] / PHE)

Hydroxyprolinemia (OH PRO)

Maple syrup urine disease (MSUD)

Alloisoleucine + Isoleucine + Leucine + Hydroxyproline (AILE + ILE + LEU + OHPRO)

Hydroxyprolinemia (OH PRO)

Maple syrup urine disease (MSUD)

Alanine + Beta Alanine + Sarcosine (ALA + BALA + SARC)

Primary lactic acidemia (various types) (LACTIC)*

Pyruvate carboxylase deficiency (PC)*

Argininosuccinate / Arginine Ratio (ASA / ARG)

Argininosuccinic aciduria (ASA)

Asparagine + Ornithine (ASN + ORN)

Girate atrophy of the retina (Hyper ORN)

Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome (HHH)*

Citrulline / Phenylalanine Ratio (CIT / PHE)

Argininosuccinic aciduria (ASA)

Carbamoyltransferase deficiency (CPS)*

Citrullinemia type I (CIT I)

Citrullinemia type II (CIT II)

Ornithine transcarbamylase deficiency (OTC)*

Pyruvate carboxylase deficiency (PC)

Methionine / Phenylalanine Ratio (MET / PHE)

Homocystinuria (HCY)

Homocystinuria-megaloblastic anemia (CBL G)

Hypermethioninemia (MET)

Methylcobalamin deficiency (CBL E)

Methylene tetrahydrofolate reductase deficiency (MTHFR)

Methylmalonic aciduria and homocystinuria (CBL C)*

Oxoproline + Pipecolate (OXOPRO + PIPA)

Pyroglutamic acidemia (OXO PRO)

Phenylalanine / Tyrosine Ratio (PHE / TYR)

Disorders of biopterin biosynthesis (BIOPT-BIO)

Disorders of biopterin regeneration (BIOPT-REG)

Hyperphenylalaninemia (variant, benign) (H-PHE)

Phenylketonuria (PKU)

Valine / Phenylalanine Ratio (VAL/PHE)

Maple syrup urine disease (MSUD)

Valinemia (Hyper VAL)

MS/MS: Acyl-Carnitine

Octenoylcarnitine (C8:1)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)*

MS/MS: Fatty Acid Oxidase

Carnitine.free (C0)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine uptake defect (CUD)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))*

Maternal carnitine uptake defect (CUD (mat))

Maternal glutaric acidemia type I (GA I (mat))

Propionic acidemia (PROP)

Decanoylcarnitine (C10)

Glutaric acidemia type II (GA II)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)*

Succinyl-CoA ligase deficiency (SUCLA2)

Decenoylcarnitine (C10:1)

Glutaric acidemia type II (GA II)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)*

Succinyl-CoA ligase deficiency (SUCLA2)

Hydroxydecenoylcarnitine (C10:1OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Maternal glutaric acidemia type I (GA I (mat))*

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)*

Trifunctional protein deficiency (TFP)*

Decadienoylcarnitine (C10:2)

2,4-Dienoyl-CoA reductase deficiency (DE RED)

Dodecanoylcarnitine (C12)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)*

Dodecenoylcarnitine (C12:1)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)*

Hydroxydodecenoylcarnitine (C12:1OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Hydroxydodecanoylcarnitine (C12OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Tetradecanoylcarnitine (C14)

Carnitine palmitoyltransferase II deficiency (CPT II)*

Carnitine-acylcarnitine translocase deficiency (CACT)*

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Tetradecenoylcarnitine (C14:1)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Hydroxytetradecenoylcarnitine (C14:1OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Tetradecadienoylcarnitine (C14:2)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Hydroxytetradecadienylcarnitine (C14:2OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Hydroxytetradecanoylcarnitine (C14OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Palmitoylcarnitine (C16)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine uptake defect (CUD)*

Carnitine-acylcarnitine translocase deficiency (CACT)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Palmitoleylcarnitine (C16:1)

Carnitine palmitoyltransferase II deficiency (CPT II)*

Carnitine-acylcarnitine translocase deficiency (CACT)*

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)*

Hydroxypalmitoleylcarnitine (C16:1OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Hydroxypalmitoylcarnitine (C16OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Stearoylcarnitine (C18)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine uptake defect (CUD)*

Carnitine-acylcarnitine translocase deficiency (CACT)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Oleylcarnitine (C18:1)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine uptake defect (CUD)*

Carnitine-acylcarnitine translocase deficiency (CACT)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Hydroxyoleylcarnitine (C18:1OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Linoleoylcarnitine (C18:2)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine uptake defect (CUD)*

Carnitine-acylcarnitine translocase deficiency (CACT)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Hydroxylinoleoylcarnitine (C18:2OH)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Hydroxybutyrylcarnitine (C4OH)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))

Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (SCHAD)

Hexanoylcarnitine (C6)

Glutaric acidemia type II (GA II)*

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)*

Hydroxyhexanoylcarnitine (C6OH)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (SCHAD)

Octanoylcarnitine (C8)

Glutaric acidemia type II (GA II)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

MS/MS: Fatty Acid Oxidase CALCULATED RATIO

Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoylcarnitine + Oleylcarnitine + Stearoylcarnitine /Citrulline Ratio ([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)

Carnitine uptake defect (CUD)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))*

Maternal carnitine uptake defect (CUD (mat))

Maternal glutaric acidemia type I (GA I (mat))

Carnitine.free / Palmitoylcarnitine+Stearoylcarnitine Ratio (C0 / [C16 + C18])

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)*

Carnitine-acylcarnitine translocase deficiency (CACT)*

Carnitine.free / Palmitoylcarnitine Ratio (C0 / C16)

Carnitine palmitoyltransferase I deficiency (CPT I)*

Carnitine.free / Stearoylcarnitine Ratio (C0 / C18)

Carnitine palmitoyltransferase I deficiency (CPT I)*

Tetradecenoylcarnitine / Dodecenoylcarnitine Ratio (C14:1 / C12:1)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)*

Tetradecenoylcarnitine / Palmitoylcarnitine Ratio (C14:1 / C16)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Tetradecenoylcarnitine / Acetylcarnitine Ratio (C14:1 / C2)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio (C16OH / C16)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Stearoylcarnitine / Propionylcarnitine Ratio (C18 / C3)

Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)*

Trifunctional protein deficiency (TFP)*

Octanoylcarnitine / Decanoylcarnitine Ratio (C8 / C10)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)*

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)*

Octanoylcarnitine / Acetylcarnitine Ratio (C8 / C2)

Glutaric acidemia type II (GA II)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

MS/MS: Fatty Acid Oxidase-Organic Acids

Acetylcarnitine (C2)

Carnitine uptake defect (CUD)

Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (SCHAD)*

MS/MS: Fatty Acid Oxidase-Organic Acids CALCULATED RATIO

Butyrylcarnitine + Isobutyrylcarnitine (C4)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isobutyrylglycinuria (IBD)

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Butyrylcarnitine + Isobutyrylcarnitine / Acetylcarnitine Ratio (C4 / C2)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isobutyrylglycinuria (IBD)

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Butyrylcarnitine + Isobutyrylcarnitine / Propionylcarnitine Ratio (C4 / C3)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isobutyrylglycinuria (IBD)

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Butyrylcarnitine + Isobutyrylcarnitine / Octanoylcarnitine Ratio (C4 / C8)

Isobutyrylglycinuria (IBD)

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Glutarylcarnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)

Glutaric acidemia type I (GA I)

Glutaric acidemia type II (GA II)

Maternal glutaric acidemia type I (GA I (mat))*

Glutarylcarnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio (C5DC + C10OH / C16)

Glutaric acidemia type I (GA I)

Glutarylcarnitine + Hydroxydecanoylcarnitine /Butyrylcarnitine + Isobutyrylcarnitine Ratio (C5DC + C10OH / C4)

Maternal glutaric acidemia type I (GA I (mat))

Glutarylcarnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio (C5DC + C10OH / C5OH)

Glutaric acidemia type I (GA I)

Glutaric acidemia type II (GA II)

Glutarylcarnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio (C5DC + C10OH / C8)

Glutaric acidemia type I (GA I)

Hydroxyoctanoylcarnitine + Malonylcarnitine (C8OH + C3DC)

Malonic acidemia (MAL)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

Hydroxyoctanoylcarnitine + Malonylcarnitine / Decanoylcarnitine Ratio (C8OH + C3DC / C10)

Malonic acidemia (MAL)

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

MS/MS: Organic Acids

Propionylcarnitine (C3)

Methylmalonic acidemia (MUT)

Methylmalonic acidemia (CBL A)

Methylmalonic aciduria and homocystinuria (CBL C)

Multiple carboxylase deficiency (MCD)*

Propionic acidemia (PROP)

Propenoylcarnitine (C3:1)

Succinyl-CoA ligase deficiency (SUCLA2)

Methylmalonylcarnitine (C4DC)

Methylmalonic acidemia (CBL A)*

Methylmalonic acidemia (MUT)*

Methylmalonic aciduria and homocystinuria (CBL C)*

Tiglylcarnitine (C5:1)

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

beta-Ketothiolase deficiency (BKT)

Ethylmalonic encephalopathy (EE)

Hydroxyisovalerylcarnitine (C5OH)

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

3-Hydroxy-3-methylglutaric aciduria (HMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)

3-Methylglutaconic aciduria (3MGA)

beta-Ketothiolase deficiency (BKT)

Maternal 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC (mat))

Multiple carboxylase deficiency (MCD)

Methylglutarylcarnitine (C6DC)

3-Hydroxy-3-methylglutaric aciduria (HMG)

Formiminoglutamic acid (FIGLU)

Ethylmalonic encephalopathy (EE)

MS/MS: Organic Acids CALCULATED RATIO

Propionylcarnitine / Carnitine.free Ratio (C3 / C0)

Methylmalonic acidemia (MUT)*

Methylmalonic acidemia (CBL A)*

Methylmalonic aciduria and homocystinuria (CBL C)*

Multiple carboxylase deficiency (MCD)*

Propionic acidemia (PROP)*

Succinyl-CoA ligase deficiency (SUCLA2)*

Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)

Formiminoglutamic acidemia (FIGLU)

Methylmalonic acidemia (CBL A)*

Methylmalonic acidemia (MUT)*

Methylmalonic aciduria and homocystinuria (CBL C)*

Multiple carboxylase deficiency (MCD)*

Propionic acidemia (PROP)*

Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Methylmalonic acidemia (MUT)

Methylmalonic acidemia (CBL A)

Methylmalonic aciduria and homocystinuria (CBL C)

Multiple carboxylase deficiency (MCD)

Propionic acidemia (PROP)

Succinyl-CoA ligase deficiency (SUCLA2)*

Propionylcarnitine / Methionine Ratio (C3 / MET)

Methylmalonic aciduria and homocystinuria (CBL C)*

Succinyl-CoA ligase deficiency (SUCLA2)*

Methylmalonylcarnitine / Hydroxyisovalerylcarnitine Ratio (C4DC / C5OH)

Maternal glutaric acidemia type I (GA I (mat))*

Methylmalonic acidemia (MUT)*

Methylmalonic acidemia (CBL A)*

Methylmalonic aciduria and homocystinuria (CBL C)*

Isovalerylcarnitine + Methylbutyrylcarnitine (C5)

2-Methylbutyrylglycinuria (2MBG)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isovaleric acidemia (IVA)

Isovalerylcarnitine + Methylbutyrylcarnitine / Carnitine.free Ratio (C5 / C0)

2-Methylbutyrylglycinuria (2MBG)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isovaleric acidemia (IVA)

Isovalerylcarnitine + Methylbutyrylcarnitine / Acetylcarnitine Ratio (C5 / C2)

2-Methylbutyrylglycinuria (2MBG)

Ethylmalonic encephalopathy (EE)

Glutaric acidemia type II (GA II)

Isovaleric acidemia (IVA)

Isovalerylcarnitine + Methylbutyrylcarnitine / Propionylcarnitine Ratio (C5 / C3)

2-Methylbutyrylglycinuria (2MBG)

Glutaric acidemia type II (GA II)

Isovaleric acidemia (IVA)

Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C50H / C8)

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

3-Hydroxy-3-methylglutaric aciduria (HMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)

3-Methylglutaconic aciduria (3MGA)

beta-Ketothiolase deficiency (BKT)

Multiple carboxylase deficiency (MCD)

Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

3-Hydroxy-3-methylglutaric aciduria (HMG)*

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)*

3-Methylglutaconic aciduria (3MGA)

beta-Ketothiolase deficiency (BKT)*

Multiple carboxylase deficiency (MCD)*