

**U.S. Department of Health and Human Services  
Office of the National Coordinator for Health Information Technology**



**Resource Guide for Newborn Screening  
Draft Detailed Use Case  
September 19, 2008**



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## Preface

The Personalized Healthcare Workgroup developed a Newborn Screening Resource 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 from 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 were approved by the American Health Information Community (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 reports that follow 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 making proper LOINC coding important for complete documentation.

Newborn screening also includes Early Hearing Detection and Intervention (EHDI) testing that is performed at the bedside in the hospital or at an ambulatory audiology testing center rather than on the newborn dried blood spot specimen. Because the results of EHDI testing are often included on the filter paper and order form used to collect the dried blood spot, or included on the birth certificate in some states, we anticipate that they may be added to single combined newborn screening test report and initial screening tests for EHDI were included in this resource guide. The focus of initial testing is only on identifying hearing loss and not the specific varieties and causes of hearing loss so only a few EHDI tests and results are included in this version of the resource guide. Future versions of the resource guide will expand to include additional data such as confirmatory hearing tests and quantitative results that help to identify the severity and type of hearing loss. There will also be tables of genetic causes of hearing loss and specific genetic alleles associated with conditions that cause hearing loss, but these are not part of the state mandated initial screening process at this 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. Future versions of the guide will also be used to store genotype information that is associated with specific phenotypes are identified by screening or confirmatory testing. This activity is just beginning and is not yet included in this version of the guide, 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 first draft of 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*<sup>1</sup>, 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.

<sup>1</sup>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.



## 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 identified by non-tandem mass spectrometry tests performed on newborn dried blood spots, as well as hearing loss detected through early hearing detection and intervention (EHD) 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.

### MS/MS: ACMG Primary Targets: Amino Acids

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

### MS/MS: ACMG Primary Targets: Fatty Acid Oxidase

| Condition [Enzyme]   | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|--|-----------|----------|-------------|-------------|
| Carnitine uptake defect<br>[Plasma membrane carnitine transporter]                         | CUD       | 212140   | TBD         | 21764004    |
| Long-chain L-3-Hydroxy dehydrogenase deficiency<br>[Long-chain L-3-Hydroxy dehydrogenase ] | LCHAD     | 609016   | 1.1.1.211   | 237999008   |
| Medium-chain acyl-CoA dehydrogenase deficiency<br>[Medium-chain acyl-CoA dehydrogenase]    | MCAD      | 607008   | 1.3.99.3    | 128596003   |
| Trifunctional protein deficiency<br>[Trifunctional protein ( $\alpha,\beta$ subunit)]      | TFP       | 609015   | 1.1.1.211   | 237999008   |



|   |       |        |           |           |
|---|-------|--------|-----------|-----------|
| Very long-chain acyl-CoA dehydrogenase deficiency<br>[Very long-chain acyl-CoA dehydrogenase] | VLCAD | 201475 | 1.3.99.13 | 237997005 |
|---|-------|--------|-----------|-----------|

## MS/MS: ACMG Primary Targets: Organic Acids

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code       | SNOMED Code |
|---|-----------|----------|-------------------|-------------|
| 3-Hydroxy-3-methylglutaric aciduria<br>[3-Hydroxy-3-methylglutaryl-CoA lyase]                               | HMG       | 300438   | 4.1.3.4           | 124611007   |
| 3-Methylcrotonyl-CoA carboxylase deficiency<br>[3-Methylcrotonyl-CoA carboxylase ( $\alpha,\beta$ subunit)] | 3MCC      | 210200   | 6.4.1.4           | 13144005    |
| Glutaric acidemia type I<br>[Glutaryl-CoA dehydrogenase]  | GA I      | 231670   | 1.3.99.7          | 76175005    |
| Isovaleric acidemia<br>[Isovaleryl-CoA dehydrogenase]   | IVA       | 243500   | 1.3.99.10         | 87827003    |
| Methylmalonic acidemia<br>[Methylmalonyl-CoA mutase]  | MUT       | TBD      | 5.4.99.2          | 42393006    |
| Methylmalonic acidemia<br>[Adenosylcobalamin synthesis]   | CBL A     | 251100   | 5.4.99.2          | 42393006    |
| Methylmalonic acidemia<br>[Adenosylcobalamin synthesis]   | CBL B     | 251110   | 5.4.99.2          | 42393006    |
| Multiple carboxylase deficiency<br>[Holocarboxylase synthetase]   | MCD       | 253270   | 6.3.4.11          | 15307001    |
| Propionic acidemia<br>[Propionyl-CoA carboxylase]   | PA        | 606054   | 6.4.1.3           | 69080001    |
| $\beta$ -Ketothiolase deficiency<br>[ $\beta$ -Ketothiolase]  | BKT       | 203750   | 2.3.1.16, 2.3.1.9 | 124265004   |

## MS/MS: ACMG Secondary Conditions: Amino Acids

| Condition [Enzyme]   | ACMG Code  | MIM Code | ENZYME Code | SNOMED Code |
|--|------------|----------|-------------|-------------|
| Argininemia<br>[Arginase]  | ARG        | 207800   | 3.5.3.1     | 23501004    |
| Citrullinemia type II<br>[Aspartate glutamate carrier (citrin)]              | CIT II     | 605814   | TBD         | TBD         |
| Disorders of biopterin biosynthesis<br>[6-Pyruvoiltetrahydropterin synthase] | BIOPT -BIO | 261640   | 4.2.3.12    | TBD         |
| Disorders of biopterin regeneration<br>[Dihydropteridine reductase]          | BIOPT-REG  | 261630   | 1.5.1.34    | 58256000    |



|  |         |        |            |           |
|--|---------|--------|------------|-----------|
| Hypermethioninemia<br>[Methionine adenosyltransferase]                 | MET     | 250850 | 2.5.1.6    | 37695001  |
| Hyperphenylalaninemia (variant, benign)<br>[Phenylalanine hydroxylase] | H-PHE   | TBD    | 1.14.16.1  | 68528007  |
| Tyrosinemia type II<br>[Tyrosine transaminase]                         | TYR II  | 276600 | 2.6.1.5    | 4887000   |
| Tyrosinemia type III<br>[4-Hydroxyphenylpyruvate acid oxidase]         | TYR III | 276710 | 1.13.11.27 | 415764005 |

## MS/MS: ACMG Secondary Conditions: Fatty Acid Oxidase

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---|-----------|----------|-------------|-------------|
| 2,4-Dienoyl-CoA reductase deficiency<br>[2,4-Dienoyl-CoA reductase]   | DE RED    | 222745   | 1.3.1.34    | TBD         |
| Carnitine acylcarnitine translocase deficiency<br>[Carnitine acylcarnitine translocase]                       | CACT      | 255110   | 2.3.1.21    | 238003000   |
| Carnitine palmitoyltransferase I deficiency<br>[Carnitine palmitoyltransferase Ia]                            | CPT I     | 255120   | 2.3.1.21    | 238001003   |
| Carnitine palmitoyltransferase II deficiency<br>[Carnitine palmitoyltransferase II]                           | CPT II    | 255110   | 2.3.1.21    | 124265004   |
| Glutaric acidemia type II<br>[ETF:ubiquinone oxidoreductase]  | GA II     | 231675   | 1.5.5.1     | 22886006    |
| Glutaric acidemia type II<br>[Electron transfer flavoprotein [ETF] ( $\alpha,\beta$ subunit)]                 | GA II     | 608053   | 1.5.5.1     | 22886006    |
| Medium-chain ketoacyl-CoA thiolase deficiency<br>[Medium-chain ketoacyl-CoA thiolase]                         | MCKAT     | 602199   | 2.3.1.16    | TBD         |
| Short-chain acyl-CoA dehydrogenase deficiency<br>[Short-chain acyl-CoA dehydrogenase]                         | SCAD      | 201470   | 1.3.99.2    | 124166007   |
| Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency<br>[Short-chain L-3-hydroxy acyl-CoA dehydrogenase] | SCHAD     | 601609   | 1.1.1.35    | 237998000   |

## MS/MS: ACMG Secondary Conditions: Organic Acids

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---|-----------|----------|-------------|-------------|
| 2-Methyl-3-hydroxybutyric aciduria<br>[2-Methyl-3-hydroxybutyryl-CoA dehydrogenase] | 2M3HBA    | 300438   | 1.1.1.178   | TBD         |
| 2-Methylbutyrylglycinuria<br>[2-Methylbutyryl-CoA dehydrogenase]                    | 2MBG      | 600301   | 1.3.99.12   | TBD         |



|   |       |        |           |           |
|---|-------|--------|-----------|-----------|
| 3-Methylglutaconic aciduria<br>[3-Methylglutaconyl-CoA hydratase]                                   | 3MGA  | 250950 | 4.2.18    | 237950009 |
| Isobutyrylglycinuria<br>[Isobutyryl-CoA dehydrogenase]  | IBD   | 604773 | 1.1.1.157 | TBD       |
| Malonic acidemia<br>[Malonyl-CoA decarboxylase]   | MAL   | 248360 | 4.1.1.9   | 361203007 |
| Methylmalonic aciduria and homocystinuria<br>[MMA mutase and homocysteine: MTHF methyl transferase] | CBL C | 277400 | 5.4.99.2  | 74653006  |
| Methylmalonic aciduria and homocystinuria<br>[MMADHC protein (brand new: see NEJM 2008;358:1454)]   | CBL D | 277410 | TBD       | TBD       |

## MS/MS: Other Conditions: Amino Acids

| Condition [Enzyme]   | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|--|-----------|----------|-------------|-------------|
| Carbamoyltransferase deficiency<br>[Carbamoyltransferase I]                                    | CPS       | 237300   | TBD         | TBD         |
| Girate atrophy of the retina<br>[Ornithine aminotransferase]                                   | Hyper ORN | 258870   | TBD         | TBD         |
| Histidinemia<br>[Histidine ammonia-lyase]  | HIS       | 235800   | TBD         | TBD         |
| Homocystinuria-megaloblastic anemia<br>[methyltetrahydrofolate homocysteine methyltransferase] | CBL G     | 250940   | TBD         | TBD         |
| Hydroxyprolinemia<br>[4-Hydroxy L-proline oxidase]   | OH PRO    | 237000   | TBD         | TBD         |
| Hyperlysinemia<br>[Lysine:α-ketoglutarate reductase]   | Hyper LYS | 238700   | TBD         | TBD         |
| Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome<br>[Ornithine translocase]         | HHH       | 238970   | TBD         | TBD         |
| Methylcobalamin deficiency<br>[Methylcobalamin]  | CBL E     | 236270   | TBD         | TBD         |
| Methylene tetrahydrofolate reductase deficiency<br>[5,10-methylene tetrahydrofolate reductase] | MTHFR     | 607093   | TBD         | TBD         |
| Nonketotic hyperglycinemia (glycine encephalopathy)<br>[Glycine cleavage system H protein]     | NKHG      | 238330   | TBD         | 237939006   |
| Ornithine transcarbamylase deficiency<br>[Ornithine transcarbamylase]                          | OTC       | 300461   | TBD         | TBD         |
| Pyroglutamic acidemia<br>[Glutathione synthetase]  | OXO PRO   | 266130   | TBD         | TBD         |



|   |           |        |     |     |
|---|-----------|--------|-----|-----|
| Pyruvate carboxylase deficiency<br>[Pyruvate carboxylase] | PC        | 266150 | TBD | TBD |
| Valinemia<br>[Valine transaminase]                        | Hyper VAL | 277100 | TBD | TBD |

### MS/MS: Other Conditions: Fatty Acid Oxidase

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---|-----------|----------|-------------|-------------|
| Maternal carnitine uptake defect<br>[Plasma membrane carnitine transporter] | CUD (mat) | 212140   | TBD         | 21764004    |

### MS/MS: Other Conditions: Organic Acids

| Condition [Enzyme]   | ACMG Code  | MIM Code | ENZYME Code | SNOMED Code |
|--|------------|----------|-------------|-------------|
| Ethylmalonic encephalopathy<br>[Unknown]   | EE         | 602473   | 1.5.5.1     | TBD         |
| Formiminoglutamic acidemia<br>[Glutamate formiminotransferase]   | FIGLU      | 229100   | TBD         | TBD         |
| Maternal 3-Methylcrotonyl-CoA carboxylase deficiency<br>[3-Methylcrotonyl-CoA carboxylase ( $\alpha,\beta$ subunit)] | 3MCC (mat) | TBD      | 6.4.1.4     | 13144005    |
| Maternal glutaric acidemia type I<br>[Glutaryl-CoA dehydrogenase]  | GA I (mat) | 231670   | 1.3.99.7    | 76175005    |
| Primary lactic acidemia (various types)<br>[MANY ENZYMES]  | LACTIC     | TBD      | TBD         | TBD         |
| Succinyl-CoA ligase deficiency<br>[Succinyl-CoA ligase, $\beta$ -subunit]  | SUCLA2     | 603921   | TBD         | TBD         |

### Non-MS/MS: Cystic Fibrosis:

| Condition [Enzyme] | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|--------------------|-----------|----------|-------------|-------------|
| Cystic fibrosis    | CF        | 602421   | N/A         | 190905008   |

### Non-MS/MS: Endocrine Disorders:

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---|-----------|----------|-------------|-------------|
| Congenital Adrenal Hyperplasia (non-classical)<br>[Steroid 21-hydroxylase deficiency] | CAH       | 201910   | 1.14.99.10  | TBD         |



|   |     |        |            |     |
|---|-----|--------|------------|-----|
| Congenital Adrenal Hyperplasia (salt-wasting)<br>[Steroid 21-hydroxylase deficiency]      | CAH | 201910 | 1.14.99.10 | TBD |
| Congenital Adrenal Hyperplasia (simple virilizing)<br>[Steroid 21-hydroxylase deficiency] | CAH | 201910 | 1.14.99.10 | TBD |
| Congenital Adrenal Hyperplasia<br>[Steroid 11-beta hydroxylase deficiency]                | CAH | 202010 | 1.14.15.4  | TBD |

### Non-MS/MS: Endocrine Disorders:

| Condition [Enzyme]                  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|-------------------------------------|-----------|----------|-------------|-------------|
| Congenital Hypothyroidism           | CH        | N/A      | N/A         | TBD         |
| Secondary Congenital Hypothyroidism | 20CH      | N/A      | N/A         | TBD         |
| Thyroid-Binding Globulin Deficiency | TBG       | 314200   | N/A         | TBD         |

### Non-MS/MS: Hemoglobin Disorders:

| Condition [Enzyme]          | ACMG Code   | MIM Code | ENZYME Code | SNOMED 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         | 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         | TBD         |
| Homozygous C disease        | FC          | N/A      | N/A         | TBD         |
| Homozygous E disease        | FE          | N/A      | N/A         | TBD         |



|                       |             |     |     |     |
|-----------------------|-------------|-----|-----|-----|
| S/Beta-thalassemia    | S/Beta-thal | N/A | N/A | TBD |
| Sickle C disease      | S/C         | N/A | N/A | TBD |
| Sickle cell anemia    | S/S         | N/A | N/A | TBD |
| Sickle D-Disease      | S/D         | N/A | N/A | TBD |
| Sickle E-Disease      | S/E         | N/A | N/A | TBD |
| Sickle O-Arab Disease | S/O Arab    | N/A | N/A | TBD |

### Non-MS/MS: Hemoglobin Disorders:

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

### Non-MS/MS: Infectious Diseases:

| Condition [Enzyme]           | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|------------------------------|-----------|----------|-------------|-------------|
| Congenital toxoplasmosis     | TOXO      | N/A      | N/A         | TBD         |
| Human immunodeficiency virus | HIV       | N/A      | N/A         | TBD         |

### Non-MS/MS: Other Disorders:

| Condition [Enzyme] | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|--------------------|-----------|----------|-------------|-------------|
|--------------------|-----------|----------|-------------|-------------|



|                          |     |     |     |     |
|--------------------------|-----|-----|-----|-----|
| Biotinidase Deficiency 2 | BIO | TBD | N/A | TBD |
|--------------------------|-----|-----|-----|-----|

### Non-MS/MS: Other Disorders:

| Condition [Enzyme]  | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---|-----------|----------|-------------|-------------|
| Classical galactosemia [galactose-1-phosphate uridylyltransferase deficiency]       | GALT      | 230400   | 2.7.7.12    | TBD         |
| Galactosepimerase deficiency [uridine diphosphate galactose 4-epimerase deficiency] | GALE4     | 230350   | 5.1.3.2     | TBD         |
| Galactokinase deficiency  | GALK4     | 230200   | 2.7.1.6     | TBD         |

### EHDI: Congenital Hearing Loss: Hearing Loss

| Condition [Enzyme]        | ACMG Code | MIM Code | ENZYME Code | SNOMED Code |
|---------------------------|-----------|----------|-------------|-------------|
| Hearing Loss, Bilateral   | HEAR      | N/A      | N/A         | TBD         |
| Hearing Loss, Left        | HEAR      | N/A      | N/A         | TBD         |
| Hearing Loss, Right       | HEAR      | N/A      | N/A         | TBD         |
| Hearing Loss, Unspecified | HEAR      | N/A      | N/A         | TBD         |



# 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 identified 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 not 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.

## 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 |
| Hydroxydeceoylcarnitine     | 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 |
| Hydroxyoleoylcarnitine      | 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 + Palmitoyl carnitine + Oleylcarnitine + Stearoylcarnitine /Citruilline 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 / Hydroxyisovalerylarnitine 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                    | C5OH / 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 |
|------------------------------|-----|-----|------------|

## 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 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 |



| Analyte  | Category | Short Name                         | LOINC Code | Units       |
|--|----------|------------------------------------|------------|-------------|
| Lysine   | AA       | LYS                                | 47689-5    | µmol/L      |
| 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 + Palmitoyl carnitine + 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      |



| Analyte   | Category | Short Name | LOINC Code | Units       |
|---|----------|------------|------------|-------------|
| Tryptophan  | AA       | TRP        | 53159-0    | µmol/L      |
| Propenoylcarnitine  | OA       | C3:1       | 53237-4    | µmol/L      |
| 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 |



| Analyte   | Category | Short Name          | LOINC Code | Units       |
|---|----------|---------------------|------------|-------------|
| Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio  | OA       | C5OH / C8           | 53172-3    | molar ratio |
| Hydroxyhexanoylcarnitine  | FAO      | C6OH                | 53173-1    | µmol/L      |
| Octenoylcarnitine   | AC       | C8:1                | 53174-9    | µmol/L      |
| 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      |



| Analyte  | Category | Short Name    | LOINC Code | Units       |
|--|----------|---------------|------------|-------------|
| Methylglutaryl carnitine                               | OA       | C6DC          | 53187-1    | µmol/L      |
| Hydroxydodecenoyl carnitine                            | FAO      | C12:1OH       | 53188-9    | µmol/L      |
| Hydroxydodecanoyl carnitine                            | FAO      | C12OH         | 53189-7    | µmol/L      |
| Tetradecadienoyl carnitine                             | FAO      | C14:2         | 53190-5    | µmol/L      |
| Tetradecenoyl carnitine                                | FAO      | C14:1         | 53191-3    | µmol/L      |
| Tetradecanoyl carnitine                                | FAO      | C14           | 53192-1    | µmol/L      |
| Tetradecenoyl carnitine / Acetylcarnitine Ratio        | FAO      | C14:1 / C2    | 53193-9    | molar ratio |
| Tetradecenoyl carnitine / Dodecenoyl carnitine Ratio   | FAO      | C14:1 / C12:1 | 53194-7    | molar ratio |
| Tetradecenoyl carnitine / Palmitoyl carnitine Ratio    | FAO      | C14:1 / C16   | 53195-4    | molar ratio |
| Hydroxytetradecadienyl carnitine                       | FAO      | C14:2OH       | 53196-2    | µmol/L      |
| Hydroxytetradecenoyl carnitine                         | FAO      | C14:1OH       | 53197-0    | µmol/L      |
| Hydroxytetradecanoyl carnitine                         | FAO      | C14OH         | 50281-5    | µmol/L      |
| Palmitoleyl carnitine                                  | FAO      | C16:1         | 53198-8    | µmol/L      |
| Palmitoyl carnitine                                    | FAO      | C16           | 53199-6    | µmol/L      |
| Argininosuccinate                                      | AA       | ASA           | 53062-6    | µmol/L      |
| Argininosuccinate / Arginine Ratio                     | AA       | ASA / ARG     | 53200-2    | molar ratio |
| Hydroxypalmitoleyl carnitine                           | FAO      | C16:1OH       | 50121-3    | µmol/L      |
| Hydroxypalmitoyl carnitine                             | FAO      | C16OH         | 50125-4    | µmol/L      |
| Hydroxypalmitoyl carnitine / Palmitoyl carnitine Ratio | FAO      | C16OH / C16   | 53201-0    | molar ratio |
| Linoleoyl carnitine                                    | FAO      | C18:2         | 45217-7    | µmol/L      |
| Oleyl carnitine  | FAO      | C18:1         | 53202-8    | µmol/L      |
| Stearoyl carnitine                                     | FAO      | C18           | 53241-6    | µmol/L      |



| Analyte                                      | Category | Short Name | LOINC Code | Units       |
|--|----------|------------|------------|-------------|
| 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      |
| 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      |



| Analyte                    | Category | Short Name | LOINC Code | Units  |
|----------------------------|----------|------------|------------|--------|
| 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 |
| Hydroxytetradecadienoylcarnitine | 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 + Palmitoyl carnitine + 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     | C5OH / 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

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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

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## MS/MS: ACMG Primary Targets: Amino Acids

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### **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 + Palmitoyl carnitine + Oleoylcarnitine + Stearoylcarnitine /Citrulline Ratio  $([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)$

Linoleoylcarnitine (C18:2)\*

Oleoylcarnitine (C18:1)\*

Palmitoylcarnitine (C16)\*

Stearoylcarnitine (C18)\*

## Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)

Hydroxydecanoylcarnitine (C10:1OH)\*

Hydroxydodecanoylcarnitine (C12OH)\*

Hydroxydodecenoylcarnitine (C12:1OH)\*

Hydroxylinoleoylcarnitine (C18:2OH)

Hydroxyoleoylcarnitine (C18:1OH)

Hydroxypalmitoleylcarnitine (C16:1OH)

Hydroxypalmitoylcarnitine (C16OH)

Hydroxypalmitoylcarnitine / Palmitoylcarnitine Ratio (C16OH / C16)

Hydroxytetradecadienylcarnitine (C14:2OH)\*

Hydroxytetradecanoylcarnitine (C14OH)

Hydroxytetradecenoylcarnitine (C14:1OH)\*

Linoleoylcarnitine (C18:2)\*

Oleoylcarnitine (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)

Hydroxydecanoylcarnitine (C10:1OH)\*  
 Hydroxydodecanoylcarnitine (C12OH)\*  
 Hydroxydodecanoylcarnitine (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)\*

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MS/MS: ACMG Primary Targets: Organic Acids

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

Hydroxyisovalerylcarnitine (C5OH)  
 Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)\*  
 Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C5OH / C8)  
 Methylglutarylcarnitine (C6DC)

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

Hydroxyisovalerylcarnitine (C5OH)  
 Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)\*  
 Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C5OH / C8)

### Glutaric acidemia type I (GA I)

Glutaryl carnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)  
 Glutaryl carnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio (C5DC + C10OH / C5OH)  
 Glutaryl carnitine + Hydroxydecanoylcarnitine / Octanoylcarnitine Ratio (C5DC + C10OH / C8)  
 Glutaryl carnitine + 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 (C5OH / C8)  
 Propionylcarnitine (C3)\*  
 Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)  
 Propionylcarnitine / Carnitine.free Ratio (C3 / C0)\*  
 Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)\*

## **Propionic acidemia (PA)**

Carnitine.free (C0)  
 Propionylcarnitine (C3)  
 Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)  
 Propionylcarnitine / Carnitine.free Ratio (C3 / C0)\*  
 Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)\*

## **β-Ketothiolase deficiency (BKT)**

Hydroxyisovalerylcarnitine (C5OH)  
 Hydroxyisovalerylcarnitine / Carnitine.free Ratio (C5OH / C0)\*  
 Hydroxyisovalerylcarnitine / Octanoylcarnitine Ratio (C5OH / C8)  
 Tiglylcarnitine (C5:1)

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MS/MS: ACMG Secondary Conditions: Amino Acids

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## **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 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)\*

## **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)\*



## 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)  
 Glutaryl carnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)  
 Glutaryl carnitine + 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)\*  
 Hydroxydecanoylcarnitine (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)

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MS/MS: ACMG Secondary Conditions: Organic Acids

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

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

## **Girata 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 (H**

UUA

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 + Palmitoyl carnitine + 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 + Palmitoyl carnitine + 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 + Palmitoyl carnitine + Oleylcarnitine + Stearoylcarnitine / Citrulline Ratio  $([C0 + C2 + C3 + C16 + C18:1 + C18] / CIT)^*$   
 Glutaryl carnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)\*  
 Glutaryl carnitine + Hydroxydecanoylcarnitine / Butyrylcarnitine + Isobutyrylcarnitine Ratio  $(C5DC + C10OH / C4)$   
 Hydroxydecanoylcarnitine (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

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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**

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MS/MS:    **Amino Acids**

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## **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)

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MS/MS:    Amino Acids    CALCULATED RATIO

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### **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 bipterin biosynthesis (BIOPT -BIO)  
 Disorders of bipterin 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 (PA)



### **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 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 dehydrogenase deficiency (LCHAD)\*  
 Trifunctional protein deficiency (TFP)\*

### **Hydroxydodecanoylcarnitine (C12OH)**

Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)\*  
 Trifunctional protein deficiency (TFP)\*

### **Tetradecanoylcarnitine (C14)**

Carnitine acylcarnitine translocase deficiency (CACT)\*  
 Carnitine palmitoyltransferase II deficiency (CPT II)\*  
 Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)



### **Tetradecenoylcarnitine (C14:1)**

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

Trifunctional protein deficiency (TFP)\*

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

### **Hydroxytetradecenoylcarnitine (C14:1OH)**

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

Trifunctional protein deficiency (TFP)\*

### **Hydroxytetradecanoylcarnitine (C14OH)**

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

Trifunctional protein deficiency (TFP)

### **Palmitoylcarnitine (C16)**

Carnitine acylcarnitine translocase deficiency (CACT)

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)

Carnitine uptake defect (CUD)\*

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

Trifunctional protein deficiency (TFP)

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

### **Palmitoleylcarnitine (C16:1)**

Carnitine acylcarnitine translocase deficiency (CACT)\*

Carnitine palmitoyltransferase II deficiency (CPT II)\*

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

Trifunctional protein deficiency (TFP)\*

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

### **Hydroxypalmitoleylcarnitine (C16:1OH)**

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

Trifunctional protein deficiency (TFP)



### **Hydroxypalmitoylcarnitine (C16OH)**

Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)  
Trifunctional protein deficiency (TFP)

### **Stearoylcarnitine (C18)**

Carnitine acylcarnitine translocase deficiency (CACT)  
Carnitine palmitoyltransferase I deficiency (CPT I)  
Carnitine palmitoyltransferase II deficiency (CPT II)  
Carnitine uptake defect (CUD)\*  
Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)\*  
Trifunctional protein deficiency (TFP)\*

### **Oleylcarnitine (C18:1)**

Carnitine acylcarnitine translocase deficiency (CACT)  
Carnitine palmitoyltransferase I deficiency (CPT I)  
Carnitine palmitoyltransferase II deficiency (CPT II)  
Carnitine uptake defect (CUD)\*  
Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)\*  
Trifunctional protein deficiency (TFP)\*

### **Hydroxyoleylcarnitine (C18:1OH)**

Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)  
Trifunctional protein deficiency (TFP)

### **Linoleoylcarnitine (C18:2)**

Carnitine acylcarnitine translocase deficiency (CACT)  
Carnitine palmitoyltransferase I deficiency (CPT I)  
Carnitine palmitoyltransferase II deficiency (CPT II)  
Carnitine uptake defect (CUD)\*  
Long-chain L-3-Hydroxy dehydrogenase deficiency (LCHAD)\*  
Trifunctional protein deficiency (TFP)\*

### **Hydroxylinoleoylcarnitine (C18:2OH)**

Long-chain L-3-Hydroxy 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)

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MS/MS: Fatty Acid Oxidase CALCULATED RATIO

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## **Carnitine.free + Acetylcarnitine + Propionylcarnitine + Palmitoyl carnitine + 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 acylcarnitine translocase deficiency (CACT)\*

Carnitine palmitoyltransferase I deficiency (CPT I)

Carnitine palmitoyltransferase II deficiency (CPT II)\*

## **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 dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

### **Stearoylcarnitine / Propionylcarnitine Ratio (C18 / C3)**

Long-chain L-3-Hydroxy 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)

### **Glutaryl carnitine + Hydroxydecanoylcarnitine (C5DC + C10OH)**

Glutaric acidemia type I (GA I)  
 Glutaric acidemia type II (GA II)  
 Maternal glutaric acidemia type I (GA I (mat))\*

### **Glutaryl carnitine + Hydroxydecanoylcarnitine / Palmitoylcarnitine Ratio (C5DC + C10OH / C16)**

Glutaric acidemia type I (GA I)

### **Glutaryl carnitine + Hydroxydecanoylcarnitine / Butyrylcarnitine + Isobutyrylcarnitine Ratio (C5DC + C10OH / C4)**

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

### **Glutaryl carnitine + Hydroxydecanoylcarnitine / Hydroxyisovalerylcarnitine Ratio (C5DC + C10OH / C5OH)**

Glutaric acidemia type I (GA I)  
 Glutaric acidemia type II (GA II)

### **Glutaryl carnitine + 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 (PA)

### 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)

Ethylmalonic encephalopathy (EE)

$\beta$ -Ketothiolase deficiency (BKT)

### Hydroxyisovalerylcarnitine (C5OH)

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

3-Hydroxy-3-methylglutaric aciduria (HMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)

3-Methylglutaconic aciduria (3MGA)

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

Multiple carboxylase deficiency (MCD)

$\beta$ -Ketothiolase deficiency (BKT)

### Methylglutarylcarnitine (C6DC)

3-Hydroxy-3-methylglutaric aciduria (HMG)



## Formiminoglutamic acid (FIGLU)

Ethylmalonic encephalopathy (EE)

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MS/MS: Organic Acids CALCULATED RATIO

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

Methylmalonic acidemia (CBL A)\*  
 Methylmalonic acidemia (MUT)\*  
 Methylmalonic aciduria and homocystinuria (CBL C)\*  
 Multiple carboxylase deficiency (MCD)\*  
 Propionic acidemia (PA)\*  
 Succinyl-CoA ligase deficiency (SUCLA2)\*

### Propionylcarnitine / Palmitoylcarnitine Ratio (C3 / C16)

Formiminoglutamic acidemia (FIGLU)  
 Methylmalonic acidemia (MUT)\*  
 Methylmalonic acidemia (CBL A)\*  
 Methylmalonic aciduria and homocystinuria (CBL C)\*  
 Multiple carboxylase deficiency (MCD)\*  
 Propionic acidemia (PA)\*

### Propionylcarnitine / Acetylcarnitine Ratio (C3 / C2)

Methylmalonic acidemia (MUT)  
 Methylmalonic acidemia (CBL A)  
 Methylmalonic aciduria and homocystinuria (CBL C)  
 Multiple carboxylase deficiency (MCD)  
 Propionic acidemia (PA)  
 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 (CBL A)\*
- Methylmalonic acidemia (MUT)\*
- 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 (C5OH / C8)**

- 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
- 3-Hydroxy-3-methylglutaric aciduria (HMG)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-Methylglutaconic aciduria (3MGA)
- Multiple carboxylase deficiency (MCD)
- $\beta$ -Ketothiolase deficiency (BKT)



## 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)

Multiple carboxylase deficiency (MCD)\*

$\beta$ -Ketothiolase deficiency (BKT)\*