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| **UAB BIOCHEMICAL GENETICS LABORATORY** | | | | | | | | |
| **720 South Twentieth Street, Room 634** **Birmingham, Alabama 35294-0024** **Phone: (205) 996-4992**  **Fax: (205) 975-2742**  [**https://www.uab.edu/medicine/genetics/clinical-laboratories/biochemical-genetics-laboratory**](https://www.uab.edu/medicine/genetics/clinical-laboratories/biochemical-genetics-laboratory) | | | **\*Overnight Specimen Mailing Address**  **The UAB Biochemical Genetics Laboratory**  **KAUL 634**  **720 20th Street South**  **Birmingham, AL 35233**  **Attn: John Moore** | | | | | |
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| **Test** | **Description** |  | | **Turn Around Time\*** | | **CPT codes** | | **Specimen Requirements** |
| **Amino Acid Analysis (ion exchange chromatography)** | | | | | | | | |
| Plasma | Quantitative analysis of plasma amino acids and related compounds. Provides diagnostic information pertaining to certain amino acidopathies, organic acidemias, and other metabolic conditions. |  | | 7 days | 82139, 82542  80502 | | A. Fasted whole blood in a green top (heparinized) tube stored at 4 C.  B. At least 1 cc plasma collected from heparinized whole blood and kept  frozen until transported to the lab. | |
| Urine | Quantitative analysis of urinary amino acids and related compounds. Provides diagnostic information pertaining to certain amino acidopathies, organic acidemias, and other metabolic conditions. |  | | 7 days | 82570, 82139 82542, 80502 | | Urine should be collected over a 12-24 hour period, pooled and kept refrigerated. At least 5 cc of the pooled sample is needed for amino acid analysis. Store frozen until transported to the lab. | |
| CSF | Quantitative analysis of CSF amino acids and related compounds. Provides diagnostic information pertaining to certain amino acidopathies, organic acidemias, and other metabolic conditions. |  | | 7 days | 82139, 82542  80502 | | A minimum of 1 cc of CSF is required for amino acid analysis. Collect on ice and store frozen. | |
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| **Organic Acid Analysis (gas chromatography/mass spectrometry)** | | | | | | | | |
| Urine | Semi-quantitative analysis of excreted organic acids and related compounds. Provides diagnostic information regarding organic acidemias, fatty acid oxidation disorders, and other conditions. |  | | 7 days | 82570, 83918, 83919, 82542, 80502 | | At least 3 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab. | |
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| **Acylcarnitine Analysis (tandem mass spectrometry)** | | | | | | | | |
| Plasma | Quantitative analysis of individual fatty acid-esterified carnitine species. Provides diagnostic information regarding fatty acid oxidation disorders and organic acidemias |  | | 7 days | 82017, 83789, 80502 | | 1-2 cc whole blood in a green top (heparinized) tube stored at 4 C or frozen plasma. | |
| **Free/Esterified Carnitine Determination (tandem mass spectrometry)** | | | | | | | | |
| Plasma | Quantitative analysis of free and esterified carnitine fractions. Complementary to acylcarnitine analysis (see below); provides diagnostic information regarding fatty acid oxidation disorders and organic acidemias. |  | | 7 days | 82379, 83789 80502 | | 1-2 cc whole blood in a green top (heparinized) tube stored at 4 C or frozen plasma. | |
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| **Comprehensive Carnitine Analysis (tandem mass spectrometry)** | | | | | | | | |
| Plasma | Combined, quantitative analysis of both free/esterified carnitine fractions and individual esterified carnitine species. Provides diagnostic information regarding fatty acid oxidation disorders and organic acidemias. |  | | 7 days | 82017,83789  82379, 80502 | | 2 cc whole blood in a green top (heparinized) tube stored at 4 C or frozen plasma. | |
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| **Phosphoethanolamine Determination (high performance ion exchange liquid chromatography)** | | | | | | | | |
| Urine | Quantitative analysis of excreted phosphoethanolamine. Provides supportive, but not diagnostic information regarding metabolic and other conditions affecting bone turnover. |  | | 7 days | 82131, 82570 82542, 80502 | | At least 5 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab. | |
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| **Methylmalonic Acid Determination (gas chromatography/mass spectrometry)** | | | | | | | | |
| Urine | Quantitative analysis of excreted methylmalonic acid. Provides diagnostic information regarding disorders of methylmalonyl-CoA and vitamin B12 (cobalamin) metabolism. |  | | 7 days | 83921, 82570, 82542, 80502 | | At least 3 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab. | |
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| **Succinylacetone Determination (gas chromatography/mass spectrometry)** | | | | | | | | |
| Urine | Quantitative analysis of excreted succinylacetone. Provides diagnostic information regarding disorders of fumarylacetoacetic acid hydrolase (FAH). |  | | 7 days | 83921, 82570, 80542, 80502 | | At least 3 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab. | |
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| **Creatine Deficiency Syndrome Analysis (liquid chromatography/tandem mass spectrometry)** | | | | | | | | |
| Urine + Plasma | Quantitative analysis of excreted and circulatory creatine, guanidinoacetate, and creatinine, which provide diagnostic information regarding disorders of the creatine metabolic pathway. |  | | 7 days | 82540(2), 82542(2), 82565, 82570, 80502 | | **Urine**: at least 3 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab.  **Plasma:** 1-2 cc plasma collected from heparinized whole blood and stored frozen until transported to the lab. | |

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| **Acylglycine analysis (mass spectrometry)** | | | | | |
| Urine | Quantitative analysis of excreted individual fatty acid-esterified glycine species. Provides diagnostic information regarding fatty acid oxidation disorders and organic acidemias |  | 7 days | 83789, 82570, 80502 | At least 3 cc of urine should be collected on ice. The first morning void is preferred. Store frozen until transported to the lab. |

\***Emergency testing is available for all services upon request (TAT < 8 hrs)**

**Please note we will continue to bill the referring laboratory, not the individual patient.**