Other Fluids

Test Name: NEUROCHEMISTRY FOR NEUROTRANSMITTER DISEASES, CSF

General Information

Lab Order Codes: CAAS (NC08) – Alpha aminoacidic semialdehyde, CSF
CSIA (NC07) – Sialic Acid, CSF
CSUCC (NC06) – Succinyladenosine, CSF
CP5P (NC05) – Pyridoxal 5’-phosphate, CSF
CTNEO (NC03) – Neopterin/Tetrahydrobiopterin, CSF
C5MET (NC01) – 5-Methyltetrahydrofolate, CSF
CPYR (MET11) – Pyruvate, CSF
CLACT (MET07) – Lactate, CSF
CAMIN (MET01) – Amino Acids, CSF
CNTM (NC04) – Neurotransmitter Metabolites, CSF
NEOP (NC02) – Neopterin, CSF

Synonyms: GTP cyclohydrolase deficiency; 6-pyruvoyl tetrahydropterin synthase deficiency; Sepiapterin reductase deficiency; Dihydropteridine reductase deficiency; Tyrosine hydroxylase deficiency; Tryptophan hydroxylase deficiency; Aromatic L-amino acid decarboxylase deficiency; Monoamine oxidase deficiency; Succinic semialdehyde dehydrogenase deficiency; Pyridoxamine phosphate oxidase deficiency; Dopamine transporter deficiency; Alpha-aminoacidic acid semialdehyde dehydrogenase deficiency

CPT Codes: CAAS (NC08): 82017 - Acylcarnitines; quantitative, each specimen
CSIA (NC07): 82017 - Acylcarnitines; quantitative, each specimen
CSUCC (NC06): 82542 - Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen
CP5P (NC05): 82542 - Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen
CTNEO (NC03): 82542 - Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen
C5MET (NC01): 82542 - Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen
CPYR (MET11): 84210 – Pyruvate
CLACT (MET07): 83605 - Lactate (lactic acid)
CAMIN (MET01): 82139 - Amino acids, 6 or more amino acids, quantitative, each specimen
CNTM (NC04): (please see 3 CPT codes listed below)
82542 – Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen
83497 – Hydroxyindolacetic acid, 5-(HIAA)
83150 – Homovanillic acid (HVA)

**NEOP (NC02): 82542** - Column chromatography, includes mass spectrometry, if performed, non-drug analytes, not elsewhere specified, qualitative or quantitative, each specimen

**Test Includes:** Quantitative results specific to the CSF test(s) listed above.

**Logistics**

**Test Indications:** Adult and pediatric neurotransmitter diseases represent a highly complex group of rare neurometabolic disorders classified on the basis of alterations in neurotransmitter metabolic pathways. Diagnosis is complicated because the measurement of metabolites in peripheral fluids is generally uninformative. CSF is required for accurate diagnosis and patient management.

CSF testing provides diagnoses for a broad range of disorders including those affecting neurotransmitter metabolism (including disorders of tetrahydrobiopterin metabolism), pyridoxine metabolism (pyridoxine responsive seizures and pyridoxamine phosphate oxidase deficiency), folate metabolism (cerebral folate deficiencies), sialic acid metabolism, purine metabolism (adenylosuccinate lyase deficiency) and disorders affecting cellular energetics.

**Lab Testing Sections:** Chemistry - Sendouts

**Referred to:** Medical Neurogenetics, Atlanta, GA
Amino Acids, CSF are performed at LabCorp (parent company of MNG)

**Phone Numbers:** MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:** Monday – Friday ONLY

**Turnaround Time:** 2 weeks
Amino Acids, CSF: 4-10 days

**Special Instructions:** See Container below. Some Neurochemistry testing requires special tubes obtained from the laboratory. Complete Medical Neurogenetics, LLC request form. Include test(s) required, sample date, date of birth, current medications and relevant history. Failure to provide this information may result in delayed testing.

**Specimen**

**Specimen Type:** CSF
<table>
<thead>
<tr>
<th>Container:</th>
<th>Test (CHC code/MNG code):</th>
<th>Conical Tube or sterile tube</th>
<th>Required Special Tubes</th>
<th>*Can be done off 5 tube kit obtained from lab with Neurotransmitter metabolites</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Amino Acid* (CAMN/MET01)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>A-amino adipic semialdehyde (CAAS/NC08)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
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<td></td>
<td>Lactate* (CLACT/MET07)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>5-Methyltetrahydrofolate (C5MET/NC01)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>Neopterin,total* (NEOP/NC02)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>Neurotransmitter Metabolites (CNTM/NC04)</td>
<td><strong>Call lab for 5 tube kit. Tube 1 required.</strong></td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>Pyridoxal 5-phosphate (CP5P/NC05)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
</tr>
<tr>
<td></td>
<td>Pyruvate (CPYR/MET11)</td>
<td>***Call lab for Perchloric Acid (7-8%) tube</td>
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<tr>
<td></td>
<td>Tetrahydrobiopterin/Neopterin Profile (CTNEO/NC03)</td>
<td><strong>Call lab for 5 tube kit. Tube 3 required</strong></td>
<td></td>
<td>YES</td>
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<tr>
<td></td>
<td>Sialic Acid (CSIA/NC07)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
</tr>
<tr>
<td></td>
<td>Succinyladenosine (CSUCC/NC06)</td>
<td>Alone</td>
<td></td>
<td>YES</td>
</tr>
</tbody>
</table>

**Draw Volume:** Dependent on tests ordered, see [Collection](#), see below

**Processed Volume:** Same as Draw Volume
<table>
<thead>
<tr>
<th>Test (CHC code/MNG code):</th>
<th>Volume (preferred)</th>
<th>Minimum Volume</th>
<th>Covered in Special 5 tube kit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amino Acid* (CAAS/NC08)</td>
<td>0.5 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>A-aminoacidic semialdehyde (CAAS/NC08)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>Lactate*(CLACT/MET07)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>5-Methyltetrahydrofolate* (C5MET/NC01)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>Neopterin,total* (NEOP/NC02)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>Neurotransmitter Metabolites** (CNTM/NC04)</td>
<td>4.5 mL (Must fill 5 tubes according to directions below). Tube 1 required.</td>
<td>0.5 mL in Tube 1 only from kit.</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>Pyruvate*** (CPYR/MET11)</td>
<td>1 mL</td>
<td>0.3 mL in special Perchloric Acid Tube</td>
<td></td>
</tr>
</tbody>
</table>

**Filling Special tubes for Neurotransmitter Metabolites:**
1. Collect CSF directly from tap needle.
2. The five tubes in the kit are numbered 1-5.
3. Fill each tube to marked line (0.5 mL in tube; 1 mL in tubes 2-5) (4.5 mL total volume CSF)
4. Attach patient identifier labels to each tube without covering the number on the tube.
5. Place tubes in biohazard bag and place on wet ice at the bedside.
6. Send to Lab immediately.

<table>
<thead>
<tr>
<th>Test (CHC code/MNG code):</th>
<th>Volume (preferred)</th>
<th>Minimum Volume</th>
<th>Covered in Special 5 tube kit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pyridoxal 5-phosphate* (CP5P/NC05)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td>OR with Neurotransmitter tubes</td>
</tr>
<tr>
<td>Test Description</td>
<td>Volume</td>
<td>Additional Information</td>
<td></td>
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<tr>
<td>---------------------------------------------------------------------------------</td>
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<td>----------------------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>Pyruvate (CPYR/MET11)</td>
<td>1 mL</td>
<td>(<strong>Must add to special tube)</strong></td>
<td></td>
</tr>
<tr>
<td>Tetrahydrobiopterin/Neopterin Profile, BH4,N* (CTNEO/NC03)</td>
<td>1 mL</td>
<td>(Must fill Tube 3 from special collection tubes)</td>
<td></td>
</tr>
<tr>
<td>Sialic Acid* (CSIA/NC07)</td>
<td>1 mL</td>
<td>0.5 mL</td>
<td></td>
</tr>
<tr>
<td>Succinyladenosine* (CSUCC/NC06)</td>
<td>1 mL</td>
<td>0.5 mL</td>
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</tbody>
</table>

**Special Processing:**

Lab Staff:
If CSF samples appear to be blood contaminated on receipt in the lab, they must be centrifuged and the spun contents transferred to conical tubes before freezing.

Store all CSF specimens in their appropriate tubes at -80°. Ship on dry ice (include 3-4 lbs in the box) Monday – Friday by overnight courier. Include requisition and list of medications and relevant clinical history with the specimens.

Note: Specimens will only be accepted Monday – Saturday and not on holidays or Sundays.

**Patient Preparation:**
None

**Sample Rejection:**
Mislabeled or unlabeled specimens; warm specimens; specimens collected inappropriately or in wrong tube

**Interpretive**

<table>
<thead>
<tr>
<th>Reference Range:</th>
<th>AGE (years)</th>
<th>0–0.2</th>
<th>0.2–0.5</th>
<th>0.5–2</th>
<th>2-5</th>
<th>5-10</th>
<th>10 - 15</th>
<th>Adults</th>
</tr>
</thead>
<tbody>
<tr>
<td>5HIAA (nmol/L)</td>
<td></td>
<td></td>
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<td></td>
<td>0-15</td>
<td>208 - 1159</td>
<td>179 - 711</td>
<td>129 - 520</td>
<td>74 - 345</td>
<td>66 - 338</td>
<td>67 - 189</td>
<td>67 - 140</td>
</tr>
<tr>
<td>HVA (nmol/L)</td>
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<tr>
<td></td>
<td>0-15</td>
<td>337 - 1299</td>
<td>450 - 1132</td>
<td>294 - 1115</td>
<td>233 - 928</td>
<td>218 - 852</td>
<td>167 - 563</td>
<td>145 - 324</td>
</tr>
<tr>
<td>Test Description</td>
<td>&lt;300</td>
<td>&lt;300</td>
<td>&lt;300</td>
<td>&lt;150</td>
<td>&lt;100</td>
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<tr>
<td>3-O-MD (nmol/L)</td>
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<tr>
<td>Neop (nmol/L)</td>
<td>7 – 65</td>
<td>7 – 65</td>
<td>7 – 65</td>
<td>7 – 65</td>
<td>7 – 40</td>
<td>8 – 33</td>
<td>8 – 28</td>
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<tr>
<td>4MTHF (nmol/L)</td>
<td>40 - 240</td>
<td>40 - 240</td>
<td>40 - 187</td>
<td>40 - 150</td>
<td>40 - 128</td>
<td>40 - 120</td>
<td>40 – 120</td>
<td></td>
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<tr>
<td>Sialic Acid, Free (umol/L)</td>
<td>4 - 55</td>
<td>4 - 55</td>
<td>4 - 22</td>
<td>4 - 22</td>
<td>4 - 22</td>
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<tr>
<td>Sialic Acid, Total (umol/L)</td>
<td>8 - 125</td>
<td>8 - 125</td>
<td>8 - 50</td>
<td>8 - 50</td>
<td>8 - 50</td>
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<tr>
<td>Sialic Acid, Ratio</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
<td>0.30 – 0.70</td>
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<tr>
<td>Lactate (mmol/dL)</td>
<td>1.11-2.44</td>
<td>1.11-2.44</td>
<td>1.11-2.44</td>
<td>1.11-2.44</td>
<td>1.11-2.44</td>
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<tr>
<td>AASA (umol/L)</td>
<td>&lt;0.1</td>
<td>&lt;0.1</td>
<td>&lt;0.1</td>
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<td>&lt;0.1</td>
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<tr>
<td>Succinyl-adenosine (umol/L)</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
<td>0.74 – 4.92</td>
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<tr>
<td>AGE (years)</td>
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<td>0 – 0.25</td>
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<td>0.26 – 1.0</td>
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<td>1.01 – 4.0</td>
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<tr>
<td>4.01 - Adult</td>
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<tr>
<td>PLP (nmol/L)</td>
<td>30 - 80</td>
<td>23 - 65</td>
<td>15 - 51</td>
<td>10 – 37</td>
<td></td>
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<tr>
<td>AGE (years)</td>
<td>0 – 30 days</td>
<td>31 days – 11 mo</td>
<td>1 yr – 17 yrs</td>
<td>Adult (&gt;17 yrs)</td>
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<tr>
<td>α-Alanine (umol/L)</td>
<td>0.0 – 1.1</td>
<td>0.0 – 1.2</td>
<td>0.0 – 1.2</td>
<td>0.0 – 2.7</td>
<td></td>
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<tr>
<td>β-Alanine (umol/L)</td>
<td>0.0 – 1.1</td>
<td>0.0 – 1.2</td>
<td>0.0 – 1.2</td>
<td>0.0 – 2.7</td>
<td></td>
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</tr>
<tr>
<td>Taurine (umol/L)</td>
<td>3.5 – 42.9</td>
<td>3.9 – 34.1</td>
<td>3.1 – 31.2</td>
<td>2.5 – 30.2</td>
<td></td>
<td></td>
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<tr>
<td>Aspartic Acid (umol/L)</td>
<td>0.0 – 17.6</td>
<td>0.0 – 13.3</td>
<td>0.0 – 12.7</td>
<td>0.0 – 14.6</td>
<td></td>
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<tr>
<td>Threonine (umol/L)</td>
<td>17.2 – 186.9</td>
<td>14.3 – 128.9</td>
<td>11.5 – 120.2</td>
<td>13.2 – 90.8</td>
<td></td>
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<tr>
<td>Serine (umol/L)</td>
<td>20.0 – 130.5</td>
<td>22.3 – 114.4</td>
<td>17.5 – 110.3</td>
<td>10.4 – 57.8</td>
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<tr>
<td>Asparagine (umol/L)</td>
<td>1.4 – 26.5</td>
<td>3.4 – 22.8</td>
<td>2.9 – 21.9</td>
<td>1.7 – 26.9</td>
<td></td>
<td></td>
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<tr>
<td>Glutamic Acid (umol/L)</td>
<td>1.3 – 61.3</td>
<td>0.0 – 58.4</td>
<td>0.0 – 104.0</td>
<td>0.0 – 84.0</td>
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<tr>
<td>Glutamine (umol/L)</td>
<td>80.7 – 1247.3</td>
<td>110.8 – 788.3</td>
<td>111.5 – 846.7</td>
<td>131.1 – 660.1</td>
<td></td>
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<tr>
<td>Glycine (umol/L)</td>
<td>2.7 – 44.5</td>
<td>2.4 – 39.7</td>
<td>2.3 – 34.1</td>
<td>2.3 – 72.7</td>
<td></td>
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</tr>
<tr>
<td>Citrulline (umol/L)</td>
<td>1.0 – 12.3</td>
<td>1.0 – 12.0</td>
<td>0.9 – 10.4</td>
<td>0.7 – 11.1</td>
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<tr>
<td>Amino Acid</td>
<td>Range 1</td>
<td>Range 2</td>
<td>Range 3</td>
<td>Range 4</td>
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<tr>
<td>Valine (umol/L)</td>
<td>10.5 – 67.6</td>
<td>10.3 – 55.3</td>
<td>8.7 – 53.3</td>
<td>6.8 – 87.2</td>
<td></td>
<td></td>
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<tr>
<td>Cystine (umol/L)</td>
<td>0.0 – 4.3</td>
<td>0.0 – 5.2</td>
<td>0.0 – 4.8</td>
<td>0.0 – 13.0</td>
<td></td>
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<tr>
<td>Methionine (umol/L)</td>
<td>1.4 – 20.4</td>
<td>1.3 – 14.7</td>
<td>1.2 – 13.5</td>
<td>1.3 – 14.2</td>
<td></td>
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</tr>
<tr>
<td>Isoleucine (umol/L)</td>
<td>2.7 – 26.4</td>
<td>2.9 – 19.7</td>
<td>2.3 – 19.1</td>
<td>1.8 – 26.3</td>
<td></td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Leucine (umol/L)</td>
<td>6.9 – 57.3</td>
<td>7.0 – 37.5</td>
<td>6.6 – 38.1</td>
<td>5.0 – 60.7</td>
<td></td>
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</tr>
<tr>
<td>Tyrosine (umol/L)</td>
<td>5.8 – 64.8</td>
<td>5.3 – 46.6</td>
<td>4.8 – 42.9</td>
<td>3.6 – 38.4</td>
<td></td>
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<tr>
<td>Phenylalanine (umol/L)</td>
<td>6.1 – 55.7</td>
<td>5.5 – 34.9</td>
<td>5.7 – 36.0</td>
<td>4.0 – 46.4</td>
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</tr>
<tr>
<td>Histadine (umol/L)</td>
<td>5.9 – 61.2</td>
<td>7.3 – 42.4</td>
<td>7.5 – 39.0</td>
<td>6.0 – 38.1</td>
<td></td>
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</tr>
<tr>
<td>GABA (umol/L)</td>
<td>0.0 – 2.1</td>
<td>0.0 – 5.7</td>
<td>0.0 – 6.2</td>
<td>0.0 – 4.3</td>
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<tr>
<td>Ornithine (umol/L)</td>
<td>1.7 – 30.3</td>
<td>2.5 – 33.0</td>
<td>1.9 – 30.2</td>
<td>1.9 – 23.6</td>
<td></td>
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</tr>
<tr>
<td>Lysine (umol/L)</td>
<td>10.7 – 71.8</td>
<td>11.2 – 60.7</td>
<td>10.3 – 57.4</td>
<td>11.5 – 84.9</td>
<td></td>
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</tr>
<tr>
<td>Arginine (umol/L)</td>
<td>4.3 – 50.3</td>
<td>8.4 – 56.3</td>
<td>8.8 – 51.9</td>
<td>9.2 – 54.7</td>
<td></td>
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</tr>
</tbody>
</table>
Methodology:  CNTM, C5MET – HPLC/Electrochemistry
NEOP, CTNEO – HPLC/Electrochemistry/fluorescence
CP5P – HPLC/fluorescence
CPYR, CLACT – Enzyme/UV Detection
CSIA, CAAS – LC/MS/MS
CSUCC – HPLC/UV
CAMIN – LC/MS-MS

References:  Medical Neurogenetics  September 2020
Phone: (678)225-0222 Fax:(678)225-0212
Lab Corp  September 2020

Updates:  1/26/2016:CPT update
11/13/2019: C4HA 4-Hydroxybutyric Acid removed, no longer offered
9/15/2020: Updated Amino Acids, CSF, now performed at Lab Corp.