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## Other Fluids

**ALab Dept:**

**Test Name: NEUROCHEMISTRY FOR NEUROTRANSMITTER DISEASES, CSF**

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### **General Information**

**Lab Order Codes:** CAAS (NC08) – Alpha amino adipic 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-pyruvoyltetrahydropterin 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-amino adipic 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.

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***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.

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***Specimen***

**Specimen Type:** CSF

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**Container:**

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

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

Same as Draw Volume

**Collection:**

Test (CHC code/MNG code):	Volume (preferred)	Minimum Volume	Covered in Special 5 tube kit
Amino Acid* (CAAS/NC08)	0.5 mL	0.5 mL	OR with Neurotransmitter tubes
A-aminoadipic semialdehyde (CAAS/NC08)	1 mL	0.5 mL	OR with Neurotransmitter tubes
Lactate*(CLACT/MET07)	1 mL	0.5 mL	OR with Neurotransmitter tubes
5-Methyltetrahydrofolate* (C5MET/NC01)	1 mL	0.5 mL	OR with Neurotransmitter tubes
Neopterin,total* (NEOP/NC02)	1 mL	0.5 mL	OR with Neurotransmitter tubes
Neurotransmitter Metabolites** (CNTM/NC04)	4.5 mL (Must fill 5 tubes according to directions below). Tube 1 required.	0.5 mL in Tube 1 only from kit.	OR with Neurotransmitter tubes
Pyruvate*** (CPYR/MET11)	1 mL	0.3 mL in special Perchloric Acid Tube	
<p><b>**Filling Special tubes for Neurotransmitter Metabolites:</b></p> <ol style="list-style-type: none"> <li>1. Collect CSF directly from tap needle.</li> <li>2. The five tubes in the kit are numbered 1-5.</li> <li>3. Fill each tube to marked line (0.5 mL in tube; 1 mL in tubes 2-5) (4.5 mL total volume CSF)</li> <li>4. Attach patient identifier labels to each tube without covering the number on the tube.</li> <li>5. Place tubes in biohazard bag and place on wet ice at the bedside.</li> <li>6. Send to Lab immediately.</li> </ol>			
Pyridoxal 5-phosphate* (CP5P/NC05)	1 mL	0.5 mL	OR with Neurotransmitter tubes

Pyruvate (CPYR/MET11)	1 mL (***Must add to special tube)		
Tetrahydrobiopterin/ Neopterin Profile, BH4,N* (CTNEO/NC03)	1 mL (Must fill Tube 3 from special collection tubes)	0.5 mL	OR with Neurotransmitter tubes
Sialic Acid* (CSIA/NC07)	1 mL	0.5 mL	OR with Neurotransmitter tubes
Succinyladenosine* (CSUCC/NC06)	1 mL	0.5 mL	OR with Neurotransmitter tubes

**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**

**Reference Range:**

AGE (years)	0–0.2	0.2–0.5	0.5–2	2-5	5-10	10 - 15	Adults
<b>5HIAA (nmol/L)</b>	208 - 1159	179 - 711	129 - 520	74 - 345	66 - 338	67 - 189	67 - 140
<b>HVA (nmol/L)</b>	337 - 1299	450 - 1132	294 - 1115	233 - 928	218 - 852	167 - 563	145 - 324

<b>3-O-MD (nmol/L)</b>	<300	<300	<300	<150	<100	<100	<100
<b>BH4 (nmol/L)</b>	40 - 105	23 - 98	18 - 58	18 - 50	9 - 40	9 - 32	10 - 30
<b>Neop (nmol/L)</b>	7 – 65	7 – 65	7 – 65	7 – 65	7 - 40	8 - 33	8 – 28
<b>4MTHF (nmol/L)</b>	40 - 240	40 - 240	40 - 187	40 - 150	40 - 128	40 - 120	40 – 120
<b>Sialic Acid, Free (umol/L)</b>	4 - 55	4 – 55	4 – 22	4 – 22	4 – 22	4 – 22	4 – 22
<b>Sialic Acid, Total (umol/L)</b>	8 - 125	8 - 125	8 - 50	8 - 50	8 - 50	8 - 50	8 - 50
<b>Sialic Acid, Ratio</b>	0.30 – 0.70	0.30 – 0.70	0.30 – 0.70	0.30 – 0.70	0.30 – 0.70	0.30 – 0.70	0.30 – 0.70
<b>Lactate (mmol/dL)</b>	1.11- 2.44	1.11- 2.44	1.11- 2.44	1.11- 2.44	1.11- 2.44	1.11- 2.44	1.11- 2.44
<b>AASA (umol/L)</b>	<0.1	<0.1	<0.1	<0.1	<0.1	<0.1	<0.1
<b>Succinyl- adenosine (umol/L)</b>	0.74 – 4.92	0.74 – 4.92	0.74 – 4.92	0.74 – 4.92	0.74 – 4.92	0.74 – 4.92	0.74 – 4.92
<b>AGE (years)</b>	<b>0 – 0.25</b>		<b>0.26 – 1.0</b>		<b>1.01 – 4.0</b>		<b>4.01 - Adult</b>
<b>PLP (nmol/L)</b>	30 - 80		23 - 65		15 - 51		10 – 37

<b>AGE (years)</b>	<b>0 – 30 days</b>	<b>31 days – 11 mo</b>	<b>1 yr – 17 yrs</b>	<b>Adult (&gt;17 yrs)</b>
<b>α-Alanine (umol/L)</b>	0.0 – 1.1	0.0 – 1.2	0.0 – 1.2	0.0 – 2.7
<b>β-Alanine (umol/L)</b>	0.0 – 1.1	0.0 – 1.2	0.0 – 1.2	0.0 – 2.7
<b>Taurine (umol/L)</b>	3.5 – 42.9	3.9 – 34.1	3.1 – 31.2	2.5 – 30.2
<b>Aspartic Acid (umol/L)</b>	0.0 – 17.6	0.0 – 13.3	0.0 – 12.7	0.0 – 14.6
<b>Threonine (umol/L)</b>	17.2 – 186.9	14.3 – 128.9	11.5 – 120.2	13.2 – 90.8
<b>Serine (umol/L)</b>	20.0 – 130.5	22.3 – 114.4	17.5 – 110.3	10.4 – 57.8
<b>Aspar- agine (umol/L)</b>	1.4 – 26.5	3.4 – 22.8	2.9 – 21.9	1.7 – 26.9
<b>Glutamic Acid (umol/L)</b>	1.3 – 61.3	0.0 – 58.4	0.0 – 104.0	0.0 – 84.0
<b>Glutamine (umol/L)</b>	80.7 – 1247.3	110.8 – 788.3	111.5 – 846.7	131.1 – 660.1
<b>Glycine (umol/L)</b>	2.7 – 44.5	2.4 – 39.7	2.3 – 34.1	2.3 – 72.7
<b>Citrulline (umol/L)</b>	1.0 – 12.3	1.0 – 12.0	0.9 – 10.4	0.7 – 11.1

<b>Valine (umol/L)</b>	10.5 – 67.6	10.3 – 55.3	8.7 – 53.3	6.8 – 87.2
<b>Cystine (umol/L)</b>	0.0 – 4.3	0.0 – 5.2	0.0 – 4.8	0.0 – 13.0
<b>Methionine (umol/L)</b>	1.4 – 20.4	1.3 – 14.7	1.2 – 13.5	1.3 – 14.2
<b>Isoleucine (umol/L)</b>	2.7 – 26.4	2.9 – 19.7	2.3 – 19.1	1.8 – 26.3
<b>Leucine (umol/L)</b>	6.9 – 57.3	7.0 – 37.5	6.6 – 38.1	5.0 – 60.7
<b>Tyrosine (umol/L)</b>	5.8 – 64.8	5.3 – 46.6	4.8 – 42.9	3.6 – 38.4
<b>Phenyl- alanine (umol/L)</b>	6.1 – 55.7	5.5 – 34.9	5.7 – 36.0	4.0 – 46.4
<b>Histadine (umol/L)</b>	5.9 – 61.2	7.3 – 42.4	7.5 – 39.0	6.0 – 38.1
<b>GABA (umol/L)</b>	0.0 – 2.1	0.0 – 5.7	0.0 – 6.2	0.0 – 4.3
<b>Ornithine (umol/L)</b>	1.7 – 30.3	2.5 – 33.0	1.9 – 30.2	1.9 – 23.6
<b>Lysine (umol/L)</b>	10.7 – 71.8	11.2 – 60.7	10.3 – 57.4	11.5 – 84.9
<b>Arginine (umol/L)</b>	4.3 – 50.3	8.4 – 56.3	8.8 – 51.9	9.2 – 54.7



**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.