

Overview

Method Name  
HPLC/Electrochemistry

NY State Available  
Yes

Specimen

Specimen Type  
CSF

Specimen Required  
Medical Neurogenetics collection kit (T657) required.  
Each collection kit contains 5 microcentrifuge tubes.

COLLECTION PROTOCOL:

- 1) CSF should be collected from the first drop into the tubes in the numbered order. Fill each tube to the marked line with the required volumes  
Tube 1: 0.5 mL  
Tube 2: 1.0 mL  
Tube 3: 1.0 mL (contains antioxidants necessary to protect the sample integrity)  
Tube 4: 1.0 mL  
Tube 5: 1.0 mL
  - If sample's not blood contaminated, the tubes should be placed on dry ice at bedside
  - If sample's are blood contaminated, the tubes should immediately be centrifuged (prior to freezing) and the clear CSF transferred to new similarly labeled tubes, then frozen
  - Store samples at -80 until they can be shipped
- 2) Complete Medical Neurogenetics, LLC request form. Include test required, sample date and date of birth.
- 3) Label tubes with patient name and ID number, leaving the tube number viewable.
- 4) Place samples inside a specimen transport bag and the Medical Neurogenetics, LLC request form inside the pouch of the transport bag.
- 5) Ship samples frozen on dry ice.

Reject Due To

Hemolysis	NA
Lipemia	NA
Icteric	NA
Other	

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
CSF	Frozen		CSF KIT

Clinical & Interpretive

Clinical Information

CSF 5-Methyltetrahydrofolate (NC01) is useful for determining a deficiency of folate in the central nervous system. CSF 5-Methyltetrahydrofolate (NC01) may also be used for assessment of Variants of Uncertain Significance (VUS) identified during genetic testing (e.g. Next Generation Sequencing or Capillary Sequencing Testing). CLINICAL 5-Methyltetrahydrofolate (5-MTHF) is the predominant form of folate in cerebrospinal fluid (CSF). Low CSF 5-MTHF levels are associated with inborn errors of metabolism affecting folate metabolism , dietary deficiency of folate, cerebral folate syndromes and Kearns-Sayre syndrome. Symptoms may include, anemia, developmental delay, seizures, depression and dementia.

Reference Values

5-Methyltetrahydrofolate

<u>Age</u>	<u>5MTHF</u>
(years)	(nmol/L)

0-0.2	40-240
0.2-0.5	40-240
0.5-2.0	40-187
2.0-5.0	40-150
5.0-10	40-128
10-15	40-120
Adults	40-120

Note: If test results are inconsistent with the clinical presentation, please call our laboratory to discuss the case and/or submit a second sample for confirmatory testing.

DISCLAIMER required by the FDA for high complexity clinical laboratories: HPLC testing was developed and its performance characteristics determined by Medical Neurogenetics. These HPLC tests have not been cleared or approved by the U.S. FDA.

Performance

PDF Report

Referral

Day(s) Performed  
Monday through Friday

Report Available  
10 to 18 days

Performing Laboratory Location  
Medical Neurogenetics, LLC

Fees & Codes

- Fees
- Authorized users can sign in to [Test Prices](#) for detailed fee information.
  - Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
  - Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

CPT Code Information  
82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
F5M	5-Methyltetrahydrofolate	47547-5

Result ID	Test Result Name	Result LOINC® Value
Z2241	5-Methyltetrahydrofolate	47547-5
Z2242	Interpretation	Not Provided