

## 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**

Age      5MTHF  
(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