

# **Test Definition: F5M**

5-Methyltetrahydrofolate

# 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 NA
Lipemia	NA
Icteric	NA NA
Other	



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# **Specimen Stability Information**

Specimen Type	Temperature	Time	Special Container
CSF	Frozen		CSF KIT

# **Clinical & Interpretive**

### **Clinical Information**

CSF 5-Methyltetrahydrofolate (NCO1) is useful for determining a deficiency of folate in the central nervous system. CSF 5-Methyltetrahydrofolate (NCO1) 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



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5-Methyltetrahydrofolate

# 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 <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

# **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