MAYO CLINIC LABORATORIES

Test Definition: FFTEN

Tetrahydrobiopterin and Neopterin Profile (BH4, N)

Overview

Method Name
HPLC/Electrochemistry/Fluorescence

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

Specimens other than	CSF in special collection kit (T657)
Anticoagulants other than	NA

Hemolysis	NA
Lipemia	NA
Icteric	NA

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
CSF	Frozen		CSF KIT

Clinical & Interpretive

Clinical Information

CSF Neopterin/Tetrahydrobiopterin (NC03) is useful for diagnosis of certain disorders of neurotransmitter metabolism. This testing 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 Tetrahydrobiopterin (BH4) serves as a cofactor for the hydroxylation of phenylalanine and in the biosynthesis of biogenic amines. Deficiency of BH4 may occur as a result of mutations causing a reduction in one of the three biosynthetic enzymes, guanosine triphosphate cyclohydrolase, 6-pyruvoyl-tetrahydropterin synthase, sepiapterin reductase, or the two regenerating enzymes, pterin-4-carbinolamine dehydratase, and dihydropteridine reductase. Defects in BH4 metabolism can result in hyperphenylalaninemia and deficiency of the neurotransmitters dopamine and serotonin. Changes in CSF neopterin may also occur in deficiency of the BH4 synthesis pathway. Disorders of BH4 metabolism are characterized by a wide range of symptoms that may include developmental delay, mental disability, behavioral disturbances, dystonia, Parkinsonian symptoms, gait disturbances, speech delay, psychomotor retardation and ptosis.

Reference Values

Tetrahydrobiopterin/Neopterin Profile

Age (years)	BH4 (nmol/L)	Neop (nmol/L)
0 – 0.2	40 – 105	7 – 65
0.2 – 0.5	23 – 98	7 – 65
0.5 – 2.0	18 – 58	7 – 65
2.0 – 5.0	18 – 50	7 – 65
5.0 – 10	9 – 40	7 – 40
10 – 15	9 – 32	8 – 33
Adults	10 – 30	8 - 28

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

An important consideration for false positive for false negative results is the improper labeling of the patient sample.

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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.

Test Classification
This test was developed and its performance characteristics determined by Medical Neurogenetics, LCC. It has not been cleared or approved by the U.S. FDA.

CPT Code Information
82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
FFTEN	Tetrahydrobiopterin/Neopterin	80168-8

Result ID	Test Result Name	Result LOINC® Value
Z2234	Neopterin	34146-1
Z2235	Tetrahydrobiopterin	59247-7
Z2236	Interpretation	80173-8