MAYO CLINIC LABORATORIES

Test Definition: FNEU

Neurotransmitter Metabolites

Overview

Method Name
HPLC-Electrochemistry

NY State Available
Yes

Specimen

Specimen Type
CSF

Specimen Required
Medical Neurogenetics collection kit (MCL Supply 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 (MCL supply T657)
Anticoagulants other than	NA
Hemolysis	NA
Lipemia	NA
Icteric	NA

Specimen Minimum Volume
4.5 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
CSF	Frozen (preferred)		CSF KIT

Clinical & Interpretive

Clinical Information

CSF Neurotransmitter Metabolites (5HIAA, HVA, 3OMD) (NC04) 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 Monoamine metabolite testing includes homovanillic acid (HVA), 3-O-methyl-Dopa (3-OMD), and 5-hydroxyindole acetic acid (5-HIAA). This test is useful in diagnosing pediatric neurotransmitter diseases affecting dopamine and serotonin metabolism in the brain. Inborn errors of metabolism and various drugs may lead to severe imbalances and disturbances in these neurotransmitter systems that are reflected by changes in the concentration of monoamines metabolites in CSF. Primary inherited defects involve deficiencies in tyrosine and tryptophan hydroxylase, aromatic amino acid decarboxylase, monoamine oxidase, dopamine beta hydroxylase and the dopamine transwporter. Other defects in the biopterin synthesis pathway may also affect dopamine and serotonin metabolism. These disorders are characterized by a wide range of symptoms that may include developmental delay, mental disability, behavioral disturbances, dystonia, seizures, encephalopathy, athetosis and ptosis.

Reference Values

Age5HIAAHVA3-O-MD

(years)(nmol/L)(nmol/L)(nmol/L)
0-0.2208-1159337-1299<300
0.2-0.5179-711450-1132<300
0.5-2.0129-520294-1115<300
2.0-5.074-345233-928<150
5.0-1066-338218-852<100
10-1567-189167-563<100
Adults67-140145-324<100
Interpretation performed by Keith Hyland, Ph.D.
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.

Performance

PDF Report

Referral

Performing Laboratory Location

Medical Neurogenetics, LLC

Fees & Codes

Test Classification

CPT Code Information

82542
83497
83150