



Patient ID SA00064611	Patient Name SAMPLEREPOR, FNEU	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00064611	Client Order Number SA00064611	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 19 Nov 2013 08:00		

Neurotransmitter Metabolites

5-Hydroxyindoleacetic acid

Y071

227 nmol/L

Homovanillic acid

Y071

585 nmol/L

3-O-methyldopa

Y071

34 nmol/L

ADDITIONAL INFORMATION

Reference Ranges:

Age (years)	5HIAA (nmol/L)	HVA (nmol/L)	3-O-MD (nmol/L)
0-0.2	208-1159	337-1299	<300
0.2-0.5	179-711	450-1132	<300
0.5-2.0	129-520	294-1115	<300
2.0-5.0	74-345	233-928	<150
5.0-10	66-338	218-852	<100
10-15	67-189	167-563	<100
Adults	67-140	145-324	<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.

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.

Interpretation:

Y071

The concentrations of neurotransmitter metabolites in cerebrospinal fluid were within our reference ranges.

Recently, a cerebral folate deficiency syndrome has been described. The clinical picture is one of developmental delay/regression, cerebellar ataxia, with or without seizures, with or without autism. This disorder is treatable with folinic acid. If your patient fits this clinical picture we can measure 5-methyltetrahydrofolate in the CSF we already have if you wish to add on this test.

Please see: Ramaekers VT, Blau N. Cerebral folate deficiency. Dev Med Child Neurol. 2004 Dec;46(12):843-51. Cerebral folate deficiency has also been described in mitochondrial disorders.

We now have biomarkers for folinic acid/pyridoxine responsive seizures (Antiquitin, ALDH7A1) that appear on our neurotransmitter metabolite chromatogram. These biomarkers were NOT seen in this patient.

Received: 20 Nov 2013 14:24

Reported: 20 Nov 2013 14:59

Performing Site Legend

Code	Laboratory	Address
Y071	Medical Neurogenetics, LLC	5424 Glenridge Drive NE, Atlanta, GA 30342