

Interpretation:

Laboratory Service Report

1-800-533-1710

Y07

| Patient Name SAMPLEREPORT,FNEU | Patient ID SA00064611 | Age 47 | Gender F | Order # SA00064611 |
|-----------------------------------|----------------------------------------------------|-----------|-------------|---------------------------|
| Ordering Phys CLIENT,CLIENT | | • | • | DOB 06/10/1966 |
| Client Order # SA00064611 | Account Information | | | Report Notes |
| Collected 11/19/2013 08:00 | C7028846-DLMP Rochester SDSC 2 - Client Support | | | |
| Printed 11/20/2013 15:03 | Rochester, MN 55901 | | | |

| Test | | Flag | Results | | Unit | Reference Value | Perform Site* |
|----------------------------|------------------------|---------------|---------|----------|--------|--------------------|------------------|
| Neurotransmitter Me | etabolites | | | | | | |
| RECEIVED: 11/20/201 | 13 14:24 REPORT | ED: 11/20/201 | 3 14:59 | | | | |
| 5-Hydroxyindol | eacetic acid | | 227 | | nmol/L | | Y07 |
| | | | | | | | 1 |
| Homovanillic a | cid | | 585 | | nmol/L | | Y07 |
| | | | | | | | 1 |
| 3-0-methyldopa | 3-0-methyldopa | | 34 | | nmol/L | | Y07 |
| Reference F | Pangeg: | | | | | | 1 |
| Age | 5HIAA | HVA | | 3-0-MD | | | |
| (years) | (nmol/L) | (nmol/I | .) | (nmol/L) | | | |
| 0-0.2 | 208-1159 | 337-129 | , | <300 | | | |
| 0.2-0.5 | 179-711 | 450-113 | | <300 | | | |
| 0.5-2.0 | 129-520 | 294-111 | .5 | <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.

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

Performing Site Legend on Last Page of Report

| Patient Name | Collection Date and Time | Report Status | |
|-------------------|--------------------------|------------------------------|--|
| SAMPLEREPORT,FNEU | 11/19/2013 08:00 | Final | |
| Page 1 of 2 | | >> Continued on Next Page >> | |

^{*} Report times for Mayo performed tests are CST/CDT



Test

Laboratory Service Report

| 1 | | 8 | 0 | 0 | -533-171 | 0 |
|---|----------|---|---|---|----------|---|
| | | | | | | |

| Patient Name SAMPLEREPORT,FNEU | Patient ID SA00064611 | Age 47 | Gender | Order # SA00064611 | |
|-----------------------------------|--------------------------|----------------------------------------------------|-------------|----------------------------|--|
| Ordering Phys | SA00004011 | 41 | <u> </u> - | DOB | |
| CLIENT,CLIENT Client Order # | Account Information | | | 06/10/1966 Report Notes | |
| SA00064611 | Account information | | | Report Notes | |
| Collected 11/19/2013 08:00 | SDSC 2 - Client Supp | C7028846-DLMP Rochester SDSC 2 - Client Support | | | |
| Printed 11/20/2013 15:03 | Rochester, MN 55901 | | | | |

Reference Perform
Flag Results Unit Value Site*

responsive seizures (Antiquitin, ALDH7A1) that appear on our neurotransmitter metabolite chromatogram. These biomarkers were NOT seen in this patient.

* Performing Site:

| | _ | | |
|------|------------------------------------------|---------------|--|
| Y071 | Medical Neurogenetics, LLC | Lab Director: | |
| 1071 | 5424 Glenridge Drive NF Atlanta GA 30342 | Lab Director: | |

| Patient Name | Collection Date and Time | Report Status | |
|-------------------|--------------------------|---------------------|--|
| SAMPLEREPORT,FNEU | 11/19/2013 08:00 | Final | |
| Page 2 of 2 | | ** End of Report ** | |

^{*} Report times for Mayo performed tests are CST/CDT