NMP NATIONAL METABOLOMICS PLATFORM

Quantitative Amino Acids SERUM

NHRPL Tariff Code:
4194
Tariff (Including VAT):
R 2,169.30*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PAAs
Turnaround time:
As a single test: 10 work days from receipt of sample at CHM. As part of a screening panel: 15 work days from receipt of sample at CHM.
Method:
Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Description

Above price includes the analysis of the sample, quantification and interpretation. Quantitative reporting for: Alanine, alpha-aminobutyric acid, asparagine, alpha-aminoadipic acid, anserine, arginine, argininosuccinic acid, beta-alanine, beta-aminoisobutyric acid, carnosine, citrulline, cystine, cystathionine, ethanolamine, glutamine, glycine, histidine, homocystine, homocitrulline, 4-hydroxyproline, isoleucine, leucine, lysine, methionine, 1-methylhystidine, 3-methylhistidine, phosphoethanolamine, proline, phenylalanine, ornithine, pipecolic acid, S-adenosylhomocysteine, sarcosine, saccharopine, serine, taurine, threonine, tyrosine, tryptophane, valine. Qualitative if requested: Sulfocysteine (marker for Molybdenum-cofactor deficiency).

Comments

1. This assay can be utilised to rule in or exclude amino acidopathies.
2. Medication intake may result in the secondary elevation of the glycine concentration.
3. Aspartic acid- and glutamic acid levels are not reported due to the unpredictibily of their stability in biological samples.
4. Protein- and blood contamination of the serum sample may result in false positive/negative findings.

Information Required with sample(s):

Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequent diagnosis of a metabolic disorder. Consent to use below information (point 4) is required according to POPIA regulation:

1. Clinical history of the patient. The referring clinician can complete the clinical history form by downloading it from the website and sending it with the sample, or emailing it to pliem@nwu.ac.za.
2. Other relevant medical reports (e.g. MRI brain, EEG, X-Ray reports, sonar reports, biopsy reports, genetic testing reports, etc) that may assist in the diagnosis of a metabolic disorder can be emailed to pliem@nwu.ac.za.
3. Cumulative, routine pathology results of the patient (including archive results available) – this must be provided and emailed to pliem@nwu.ac.za by the referring pathology laboratory.
4. Please download and complete the short consent form from the website and also indicate if the patient/family may be contacted by our rare disease biobank.

Sample requirements, viability, stability:

1. Samples for serum amino acid analysis should ideally be collected after an overnight fast. In neonates and infants, fasting is not required; where practical, samples may be collected immediately prior to the next scheduled feed. Sample collection should not be delayed in cases where an inherited metabolic disease is suspected, and testing may be performed regardless of feeding status.
2. A minimum of 1 ml SST (yellow top tube) serum is required.
3. Sample should be frozen, and kept frozen during transport.
4. A haemolysed sample is NOT viable for testing as this may lead to the reporting of false positive / negative findings, specifically with regards to ornithine- and arginine levels.
5. Viability: 6 Months kept frozen