Centre for Human Metabolomics (CHM)

Test:	**Quantitative Amino Acids URINE**
CHM LAB Mnemonic:	**PAAu**
NHRPL Tariff code:	4221 + 4321 + 4188 + 4194
Tariff (including VAT):	R 1 770,14
Description:	**Above price includes the assay, 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, histidine, homocystine, homocitrulline, 4-hydroxyproline, isoleucine, leucine, lysine, methionine, 1-methylhystidine, 3-methylhistidine, phosphoethanolamine, phosphoserine, proline, phenylalanine, ornithine, pipecolic acid, S-adenosylhomocysteine, sarcosine, saccharopine, serine, taurine, threonine, tyrosine, tryptophane, valine. **Qualitative if requested:** Sulfocysteine (marker for Molybdenum-cofactor deficiency)
Turnaround time:	1. Single assay: 14 workdays from receipt of sample at our laboratory 2. Part of full metabolic evaluation: 20 work days from receipt of sample at our laboratory
Comments:	1. This test is informative with regards to amino acid transporter related disorders as well as supportive profiling for amino acidopathies. 2. Medication intake may result in the secondary elevation of glycine concentration. 3. Aspartic acid and glutamic acid levels are not reported due to the unpredictibily of their stability in biological samples. 4. Bacterial, protein and blood contamination of the urine sample may result in false positive/negative findings.
Sample requirements, viability, stability:	1. **2 ml urine**, NO preservatives added, frozen overnight, send on dry ice. 2. Viability: 1 year – kept frozen
Information Required with sample(s):	Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequen 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 on our website at https://pliem.co.za/test-request-form OR download the clinical history form from our website (same link) and send it with sample/email 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) which 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 complete the short consent form (https://pliem.co.za/test-request-form) and also indicate if the patient/family would like to be contacted by our Rare Disease Biobank.
Method:	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
Reference range & units:	Reference ranges – age dependant & available upon request. Units: mmol/mol creatinine.
Contact no for results & other enquiries:	018 299 2312 (Call centre): 1) Result, patient, sample and TAT inquiries, 2) Diagnostic/interpretation services, 3) Biobank inquiries
E-mail address:	pliem@nwu.ac.za
Delivery address for samples:	Centre for Human Metabolomics (CHM), Sample reception laboratory (all sites) 11 Hoffmann Street, Building F3, Lab Number G19 (new building ground floor) North West University (NWU), Potchefstroom, 2531

PLEASE NOTE: Collection, courier and administration costs are not included.
Protocol for each individual test is available on our website: www.pliem.co.za
Valid: 1 January 2022 - 31 December 2022