NMP NATIONAL METABOLOMICS PLATFORM

Quantitative Amino Acids URINE

NHRPL Tariff Code:
4221 + 4321 + 4188 + 4194
Tariff (Including VAT):
R 2,341.92*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PAAu & PCULu (creat, u/acid, labstix)
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 pre-analytical assessment of the sample (labstix, creatinine and uric acid) and subsequent analysis, 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 test is informative with regards to amino acid transporter related disorders including cystinuria 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.
5. Labstix, creatinine and uric acid included in all urine referrals.

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. In stable adults and older children, samples for urine amino acid analysis should ideally be collected after an overnight fast. In neonates and infants, fasting is not required and random urine samples are acceptable. Sample collection should not be delayed where an inherited metabolic disease is suspected.
2. A minimum of 2 ml random urine is required for total amino acid profile
3. Sample should be frozen, and kept frozen during transport.
4. Viability: 12 months – kept frozen.
5. Sample should not be acidified and no preservatives should be added. The former affects the LC-MS/MS base analysis