NMP NATIONAL METABOLOMICS PLATFORM

Full Metabolic Evaluation
URINE

NHRPL Tariff Code:
4221 + 4321 + 4188 + 4248 + 4268 + (4285 x2) + 4216 + 4326 + 4194 + (4020 x 2) + 4022
Tariff (Including VAT):
R 9,297.42*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PMSUR2024
Turnaround time:
15 work days from receipt of sample at CHM
Method:
Pre-analytical screen / GC-MS / LC-MS/MS / Spectrophotometric / Thin-layer chromotography

Description

Biochemical analyses, quantification/qualification and interpretation: U-Creatinine, U-Uric Acid, U-Labstix, U-Reducing substances, QU-Organic acids, U-TLC-Oligosaccharides, U-MPS-DMB-screen, U-MPS GAGs LCMS/MS, QU-Amino Acids, QU-Carnitine profile.

Comments

1. This test can be utilised to rule in or exclude: amino acidurias, organic acidemias / acidurias, some fatty acid oxidation disorders, carbohydrate related disorders as well as mucopolysaccharidosis. Adding serum amino acids and acylcarnitines to the profile may be more informative with regards to amino acidopathies, carnitine transporter related disorders as well as the full spectrum of fatty acid oxidation disorders. The full profile does not exclude all known inborn errors of metabolic disorders, as biomarkers for some may be limited.
2. Medication intake/diet may significantly influence the analysis and subsequent result interpretation.
3. Bacterial- and blood contamination of the urine 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. A minimum of 10-15 ml random urine is required.
2. Sample should be frozen, and kept frozen during transport.
3. Viability: 12 months – kept frozen. Succinylacetone stability: no more than 7 days.
4. NO preservatives should be added.