Quantitative Organic Acids (Includes orotic acid, includes succinylacetone on request) URINE

NHRPL Tariff Code:
4221 + 4321 + 4188 + 4268
Tariff (Including VAT):
R 3421.47*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
POAu & 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:
Gas chromatography-mass spectrometry (GC-MS)

Description

Above price includes the following:

1. Assay, quantification and interpretation.
2. Labstix, creatinine and uric acid included in all urine referrals.

Comments

1. This test can be utilised in the diagnosis of an organic aciduria/acidemia and to evaluate if secondary markers, associated with fatty acid oxidation disorders, are present.
2. Medication intake/diet may significantly influence the analysis and subsequent result interpretation.
3. Bacterial-, protein- 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 5-7 ml random urine is required. If succinylacetone is requested, the container should be protected from light (cover in foil) and paperwork should indicate for succinylacetone analysis.
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.