Quantitative Acylcarnitines SERUM

NHRPL Tariff Code:
4020 x 2 (Free carnitine) 4022 x 1 (Acylcarnitine)
Tariff (Including VAT):
R 1,486.19*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PCARNb
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:
Tandem Mass-spectrometry

Description

Above price includes the assay, quantification and interpretation

Comments

1. This test is informative with regards to the presence of carnitine transporter related disorders, fatty acid oxidation disorders and is supportive in the diagnosis of organic acidemias.
2. Medication intake may significantly influence the analysis and subsequent result interpretation.
3. 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. A minimum of 1 ml SST (yellow top tube) serum (pre-prandial) is required.
2. Sample should be frozen, and kept frozen during transport.
3. A haemolysed sample is NOT viable for testing as this may lead to the reporting of falsely elevated long-chain acylcarnitine.
4. Viability: 6 Months kept frozen