Quantitative Very Long Chain Fatty Acids, Phytanic acid, Pristanic acid SERUM

NHRPL Tariff Code:
4268 x 2
Tariff (Including VAT):
R 3248.84*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PPEROXb
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 assay, quantification (C22, C24, C26, pristanic acid, phytanic acid) and interpretation.

Comments

1. This assay is utilised in the diagnosis of peroxisomal disorders.
2. The intake of peanut butter or a ketogenic diet should be avoided for 48 hours (or longer) before sample collection.

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 fasting sample is required.
2. Other sample requirements: 2 ml SST (yellow top tube) serum required. Sample must be separated, transferred into another tube, and kept frozen overnight.
3. Sample collection for patients 3.1) below 18 months of age / 3.2) clinically unstable / 3.3) poor fasting tolerances: should proceed 3-4 hours AFTER feeding / meal.
4. Viability: 6 months – kept frozen.