Trimethylaminuria (TMAU): Quantitative TMA (trimethylamine) and FMO3 gene sequencing URINE + EDTA BLOOD

NHRPL Tariff Code:
4268 x 2 (Urine analysis)
4268 x 2 (DNA analysis)
Tariff (Including VAT):
R 6,497.67*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PTMAu & PTMAdna
Turnaround time:
These tests are typically performed twice per year due to the low number of requests for TMAU testing and to minimise associated costs. Please contact the laboratory if you plan to request this test. The turnaround time is one month (business days, excluding weekends and public holidays) for TMA urine analysis and sequencing of the FMO3 gene, calculated from receipt of the loading test sample(s) at NMP.

Description

Above price includes the assay, quantification and interpretation

Sample requirements

1. No preservatives or acidification should be added to the urine samples.
2. No random sample without TMA loading will be tested to avoid false negative results.
3. TMA loading is a requirement for this assay – additional information on the loading protocol and collection procedure can be found here

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.
5. Due to the cost involved, the following two factors should be investigated and excluded before a TMAu loading test (urine and genetic testing) is considered. Please forward the relevant results to us once available so that they can be placed in the patient's file.

a) Exclude Helicobacter pylori infection; and
b) In cases of halitosis, consult a dentist to assess for the presence of plaque on the posterior dorsal surface of the tongue.