Quantitative Glycine Specific Assay
CSF + SERUM

NHRPL Tariff Code:
4194 – Serum Glycine
4194 – CSF Glycine
Tariff (Including VAT):
R 4,338.60*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PAA (S) & PGLYcsf & PGLYratio
Turnaround time:
15 work days from receipt of sample at CHM.
Method:
Liquid chromatography Tandemmass spectrometry (LC-MS/MS). Note: Method is validated for serum but not for CSF (as sample matrix).

Description

Above price includes the assay, quantification and interpretation

Comments

1. This assay is utilised to evaluate if non-ketotic hyperglycinemia (NKHG) is present/absent.
2. Medication (mostly anti-convulsant) intake may significantly influence the analysis and subsequent result interpretation.
3. Blood contaminated CSF sample is NOT viable for testing as this may lead to a false positive diagnosis. Haemolysis of the serum may to some extent influence the result interpretation and should be avoided.

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:

Serum + CSF required (same sample date)
1. The Quantitative Amino Acids (Serum) test will be used to analyse serum glycine. Please follow the procedure provided.
2. 1 ml CSF (cerebrospinal fluid) sample, kept frozen.
3. Viability: 6 months – kept frozen