Quantitative Galactose-1-Phosphate DRIED BLOOD SPOT

NHRPL Tariff Code:
4238
Tariff (Including VAT):
R 766.49*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
NPGAL
Turnaround time:
1 Month from receipt of sample at CHM.
Analyses take place on the second Tuesday of every month.
Method:
Tandem-Mass Spectrometry

Description

Above price includes the assay, quantification and interpretation

Comments

This test is used primarily for monitoring of galactosemia patients on treatment. It can also be used diagnostically, provided that the patient is not on a lactose free diet. It is particularly useful when a patient has received a recent blood transfusion.

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. 1x Dried blood spot [DBS] sample – 4 complete circles.
2. Keep in sealed paper envelope after dried according to requirements, send separate from other wet specimens and within 2 days after collection. Humidity and extreme temperature may influence the stability of metabolites.
3. Viability: 1 month, kept in a dry, cool place.