

Centre for Human Metabolomics (CHM)

Test:	Quantitative Galactose-1-Phosphate BLOOD CARD SAMPLE [DBS]
CHM LAB Mnemonic:	NPGAL
NHRPL Tariff code:	4238
Tariff (including VAT):	R 579,36
Description:	Above price includes the assay, quantification and interpretation
Turnaround time:	10 work days from receipt of sample at our laboratory
Comments:	<ol style="list-style-type: none"> 1. This test can be utilised in the diagnosis of galactosemia due to GALT deficiency OR can be used as marker in treatment monitoring. 2. Suspected GALT patients already on a lactose/galactose free diet may have normal galactose-1-phosphate levels. In this instance it cannot be used in 1st line diagnostics.
Sample requirements, viability, stability:	<ol style="list-style-type: none"> 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.
Information Required with sample(s):	<p>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.</p> <ol style="list-style-type: none"> 1. Clinical history of the patient. The referring clinician can complete the clinical history form on our website at https://pliem.co.za/test-request-form OR download the clinical history form from our website (same link) and send it with sample/email 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) which 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 complete the short consent form (https://pliem.co.za/test-request-form) and also indicate if the patient/family would like to be contacted by our Rare Disease Biobank.
Method:	Tandem-Mass Spectrometry
Reference ranges & units:	Reference ranges is dependent on undiagnosed vs diagnosed patients on therapeutic intervention and could be provided upon request Units: mmol/l
Contact no for results & other enquiries:	018 299 2312 (Call centre): 1) Result, patient, sample and TAT inquiries, 2) Diagnostic/interpretation services, 3) Biobank inquiries
E-mail address:	pliem@nwu.ac.za
Delivery address for samples:	Centre for Human Metabolomics (CHM), Sample reception laboratory (all sites) 11 Hoffmann Street, Building F3, Lab Number G19 (new building ground floor) North West University (NWU), Potchefstroom, 2531