Centre for Human Metabolomics (CHM)

Test:	**Glycoprotein (transferrin) analysis to screen for congenital disorder of glycosylation (CDG)- Serum AND Dried blood spot (DBS) sample**
CHM LAB Mnemonic:	**PCDGg**
NHRPL Tariff code:	4268
Tariff (including VAT):	R 2 455,63
Description:	Above price includes the assay, quantification and interpretation
Turnaround time:	30 work days from receipt of sample at our laboratory
Comments:	1. This test is utilised in the diagnosis of **congential disorders of glycosylation**. This include **type 1** and **to some extent type 2 subtypes**. Some CDGs (such as ALG13-CDG) do not present with an abnormal glycoprotein profile. 2. Blood transfussion may influence the analysis. We recommend sample collection for this test, 2 weeks after transfussion, to prevent false negative results.
Sample requirements, viability, stability:	1. **VERY IMPORTANT: 2ml SST serum sample** AND **1 x Dried blood spot (DBS) sample**. If **only one sample type is available, it should always be a serum sample. Unfortunately no testing will be done if only a DBS is received.** 2. **Serum sample**: spin sample(s) down, separate serum, transfer serum to another tube, freeze overnight, send on dry ice. 3. **DBS sample**: 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. 4. Viability: Serum - 6 months. DBS: 1 month, kept in a dry, cool place.
Information Required with sample(s):	Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequen 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 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:	LC-MS/MS glycoprotein (transferrin) analysis - Newly implemented from 1 Jan 2022
Reference range & units:	Reference ranges and units not applicable, as pattern recognition is utilised for CDG typing.
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

PLEASE NOTE: Collection, courier and administration costs are not included.
Protocol for each individual test is available on our website: www.pliem.co.za
Valid: 1 January 2022 - 31 December 2022