

Centre for Human Metabolomics (CHM)

Test:	CDG-screening (congenital disorder of glycosylation / carbohydrate deficient glycoprotein) BLOOD CARD SAMPLE [DBS]
PLIEM Mnemonic:	PCDGg
NHRPL Tariff code:	4268
Tariff (including VAT):	R 2,343.16
Description:	Assay and Interpretation
Turnaround time:	30 work days from receipt of sample at our laboratory
Transit stability / Sample viability:	Keep in sealed paper envelope, after dried according to requirements, send separate from other specimens. Viability: 1 month kept in a dry place.
Comments:	<ol style="list-style-type: none"> 1. Place dried blood card [DBS] in sealed paper envelope and NOT in plastic bag. 2. Blood transfusion may influence the analysis. We recommend this test 2 weeks after transfusion to prevent false negative. 3. Blood card [DBS] must NOT be placed in envelope before completely dry.
Sample required:	<ol style="list-style-type: none"> 1. 1 x Blood collection card (Guthrie card / DBS) by heel prick (<1 year) of age OR fingerstick (> 1 year of age). 2. Allow blood to dry on the filter paper at ambient temperature in a horizontal position for 3 hours. Required: Whatman Protein Saver 903 Paper. 3. If blood is not applied directly onto the filter paper, do not use EDTA or citrate tubes or capillaries to collect the blood.
Information Required with sample(s):	<p>Absent clinical details may affect the interpretation of results and recommendations for further/additional testing (to assist with a differential diagnosis) cannot be made.</p> <ol style="list-style-type: none"> 1. Clinical history of the patient. The referring clinician could complete and submit the clinical history on our website at https://pliem.co.za/test-request-form OR download the clinical history form from our website (same link) and email the completed form back to our laboratory at ansie.mienie@nwu.ac.za / pliem@nwu.ac.za. 2. Other significant medical reports for the patient (e.g. MRI brain, EEG, X-Ray reports, sonar reports, biopsy reports, genetic testing reports, etc). The referring clinician must please email these additional reports to ansie.mienie@nwu.ac.za. 3. Cumulative, routine pathology results of the patient (including archive results available) - this must be provided to our laboratory by the referring pathology laboratory. It could be e-mailed to pliem@nwu.ac.za OR send together with the sample(s) of the patient.
Method:	Isoelectric focusing (IEF) of transferrin proteins
Reference range & units:	Not applicable
Contact for results & other enquiries:	Sample reception and resulting
Telephone number:	018 299 2312 / 018 285 2652 (leave message)
Fax number:	018 299 2316
E-mail address:	pliem@nwu.ac.za
Delivery address for samples:	Center for Human Metabolomics (CHM), Sample reception (PLIEM/NBS/CRS) Building F3, Room Number G19, 11 Hoffmann street North West University, Potchefstroom, 2531