

Centre for Human Metabolomics (CHM)

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| Test: | Biotinidase Enzyme Activity Determination - BLOOD CARD SAMPLE [DBS] |
| PLIEM Mnemonic: | NPBIOT |
| NHRPL Tariff code: | 4268 |
| Tariff (including VAT): | R 2,190.18 |
| Description: | Assay, quantification and interpretation |
| Turnaround time: | 15 work days from receipt of sample at PLIEM laboratory |
| Transit stability / Sample viability: | Keep in sealed envelope after dried according to requirements, send separate from other specimens and within 2 days after collection. Viability: 1 month, kept in a dry place. |
| Comments: | 1. Place dried blood card [DBS] in sealed paper envelope and NOT in plastic bag. 2. Blood card [DBS] must NOT be placed in envelope before completely dry. |
| Sample required: | 1x Blood Card Sample [DBS] – 4 complete circles |
| Information Required with sample(s): | Absent clinical details may affect the interpretation of results and recommendations for further/additional testing (to assist with a differential diagnosis) cannot be made. 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: | Biotinidase assay kit: PERKIN ELMER |
| Reference range & units: | Given in percentages (%) of biotinidase enzyme activity: < 10% - biotinidase deficiency 10-30% - partial biotinidase deficiency > 30% - normal |
| Contact for results & other inquiries: | 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 |