

Centre for Human Metabolomics (CHM)

Test:	Biotinidase Enzyme Activity Determination - BLOOD CARD SAMPLE [DBS]
CHM LAB Mnemonic:	NPBIOT
NHRPL Tariff code:	4268
Tariff (including VAT):	R1,133.42
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 is utilised in the diagnosis of profound or partial biotinidase deficiency 2. Blood transfusion may influence the analysis. We recommend sample collection for this test, at least 72hrs after the transfusion, to avoid false negative findings.
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 the biotinidase activity 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. <u>Consent to use below information (point 4) is required according to POPIA regulation.</u></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:	Biotinidase assay kit: PERKIN ELMER
Reference range & units:	Biotinidase enzyme activity is expressed in % in relation to normal activity. Reference range can be provided upon request
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