

Centre for Human Metabolomics (CHM)

Test:	Mitochondrial respiratory chain enzyme analyses (muscle)(PRIVATE)
CHM LAB Mnemonic:	MITO2018
NHRPL Tariff code:	4256 x 6 (6 enzymes) + 4117 x 1 (protein assay)
Tariff (including VAT):	R 11 143,74
Description:	Above price includes the assay, quantification and and results interpretation for complexes I, II, II+III, III, IV and citrate synthase.
Turnaround time:	35-60 work days from receipt of sample at our laboratory
Comments:	<ol style="list-style-type: none"> 1. This analysis is utilised to rule in or evaluate if mitochondrial Complex I, II, Combined II+III, II or IV may be present. 2. Providing the blood lactate, lactate/pyruvate ratio result (performed at routine path lab) is advised when submitting the sample for testing.
Sample requirements, viability, stability:	<ol style="list-style-type: none"> 1. A minimum of 100 mg skeletal muscle sample (preferably from vastus lateralis) is required (size: ± half the size of a ten cent coin). 2. The biopsy shall be placed as is (without any additional preservatives or liquids) in a 1.5ml eppendorf tube (eppie) on dry ice, IMMEDIATELY after the muscle was collected. 3. Freeze the tube with sample immediately after collection at -80°C. The sample shall be stored at -80°C until it is transported to our laboratory on dry ice. 4. The samples shall be shipped on dry ice, early in the week to prevent weekend delays. 5. Note: Samples will not be analysed if above protocol is not followed.
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:	Spectrophotometric assay
Reference range & units:	Units: nmol/min/UCS (UCS = µmol/min citrate synthase activity). Information with regards to reference ranges will be provided with profile.
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:	URGENT: Muscle biopsy sample on dry ice. 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