Mitochondrial respiratory chain enzyme analyses (NHLS)
MUSCLE

NHRPL Tariff Code:
4256 x 6 (6 enzymes) + 4117 x 1 (protein assay)
Tariff (Including VAT):
R 6153.57* (Tariff only applicable to NHLS-referrals)
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PMITOnhls
Turnaround time:
35-60 work days from receipt of sample at CHM
Method:
Spectrophotometric assay

Description

Above price includes the assay, quantification and result interpretation for complexes I, II, II+III, III, IV and citrate synthase.

Comments

1. This analysis is utilized to rule in or evaluate if mitochondrial complex I, II, combined II+III, III or IV may be present.
2. Providing the blood lactate, lactate/pyruvate ratio result (performed at routine pathology laboratories) is advised when submitting the sample for testing.

Information Required with sample(s):

Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequent 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 by downloading it from the website and sending it with the sample, or emailing 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) that 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 download and complete the short consent form from the website and also indicate if the patient/family may be contacted by our rare disease biobank.

Sample requirements, viability, stability:

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 must be placed as is (without any additional preservatives or liquids) in a 1.5ml Eppendorf tube (eppie).
3. Freeze the tube with sample immediately after collection at -80°C. The sample should be stored at -80°C until it can be transported to our laboratory.
4. Must be shipped frozen on dry ice.
5. Important: Samples will not be analyzed if above protocol is not followed.