NMP NATIONAL METABOLOMICS PLATFORM

Selective Lysosomal disorder screening (PLSD) DRIED BLOOD SPOT

NHRPL Tariff Code:
N/A
Tariff (Including VAT):
Industry sponsored test. DBS-test-kits are available at all routine pathology laboratories.
Mnemonic:
Sponsored
Turnaround time:
Ten work days from receipt of sample at CHM for the 6-plex-tests. Eight to twelve weeks for international referrals.
Method:
Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Description

Quantitative enzyme activity analysis of acid beta-glucosidase (GBA), acid alpha-glucosidase (GAA), Sphingomyelinase (SMPD1), alpha-galactosidase (GLA), galactocerebrosidase (GALC), alpha-L-iduronidase (IDUA). The test also include the following reflex international testing based on the following clinical criteria that must be provided:

1. Gaucher disease suspected and Gaucher/ASMD is selected on the request form: International referral for lyso-GL1/Gb1 testing.
2. Mucopolysaccharidosis suspected and MPS panel is selected on the request form: International referral for expanded mucopolysaccharidosis testing including MPS II, IIIb, IVa, VI and VII.
3. Fabry disease suspected in a female patient and Fabry disease is selected on the request form: International referral for Biomarker Lyso-GL3/Gb3 testing.
4. International referral for genetic testing if enzyme activity testing is suggestive of Gaucher, Niemann-Pick A/B, Pompe, Fabry or MPS I, II, IIIb, IVa, VI or VII. Krabbe disease is not included.
5. International referral lyso-GL1/Gb1, -GL3/Gb3 and -ASM measurement for monitoring of patients previously diagnosed with Gaucher, Fabry and Niemann-Pick A/B respectively.

Please note that selecting all options or none on the DBS-card will result in NO reflex testing

Comments

The 6-Plex-test and associated reflex referrals can be used for the diagnosis of the following defects:
– Gaucher-, Niemann-Pick A/B-, Pompe-, Fabry- and Krabbe-disease.
– MPS I (Hurler-Scheie), II (Hunter), IIIb (Sanfilippo), IVa (Morquio), VI MPS VI (Maroteaux-Lamy), VII (Sly).
Genetic confirmation is provided for all except Krabbe disease.

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. All samples must be collected using DBS-test-kits provided by the CHM to routine pathology laboratories.
2. Sample collection should be done according to the instructions provided in the kit: EDTA samples should NOT be used to spot the blood on the DBS-card.
3. Collection method: heelprick for children younger than 1 year of age & fingerprick for patients older than 1 year of age.
4. DBS-samples must be referred on a separate pathology laboratory requisition number and should not include any other requests/samples.
5. Allow blood to dry on the filter paper at ambient temperature in a horizontal position for at least 4 hours. Avoid exposing DBS-samples to extreme heat OR direct sunlight.