Centre for Human Metabolomics (CHM)

Test:	**Selective Lysosomal disorder screening [PLSD] - BLOOD SPOT SAMPLES [DBS]**
PLIEM Mnemonic:	**PPLSDg**
Tariff (including VAT):	Sponsored testing
Description:	Assay(s), quantification and interpretation of selective lysosomal enzymes (6-plex-test) results. An expanded MPS enzyme panel, Lyso-GL1 and Lyso-GL3 testing are available upon request and performed at an external international laboratory.
Turnaround time:	1. The basic 6-plex-test result will be available 10 work days from receipt of the sample at our laboratory (excluding public holidays and weekends). 2. Expanded MPS enzyme panel (MPS II, IIIB, IVA, VI, VII, Lyso-GL3, Lyso-GL1 are performed at an external lab in Germany (TAT: 1-2 months). Abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease and should be confirmed with genetic testing which may take up to 3 months (referal to an external laboratory).
Comments:	The above test can be utilised to rule in or exclude the following disorders on enzymatic- and subsequent genetic level. These include: Fabry disease (α-galactosidase def), Krabbe disease (galactocerebrosidase def), Gaucher disease (ß-glucosidase def), Niemann-Pick disease A / B (sphingomyelinase def), MPS I (α-L-iduronidase def), Pompe / Glycogen storage defect type II (α-glucosidase def). Lyso-GL1 substrate accumulation for Gaucher's disease and Lyso-GL3 substrate accumulation in female Fabry patients can be performed upon request.
Sample requirements, viability, stability:	1. 1 x Dried blood spot (DBS) sample collected by **heal prick (younger than 6 months of age)** OR **finger prick (older than 6 months of age)** is required. 2. DBS sample collection shall only be done on the **specialised DBS-cards** for the project collaboration between **CHM, NWU, Potchefstroom** and **Sanofi/Genzyme**. **CHM, NWU, Potchefstroom shall be contacted for DBS-collection-kits ordering (no charge), logistics and new procedure as handled by our laboratory as from 1 January 2022 (piem@nwu.ac.za / ansie.mienie@nwu.ac.za)**. 2. Allow blood to **dry** on the filter paper at ambient temperature in a horizontal position for **at least 3-4 hours.** 3. If blood is not applied directly onto the filter paper, do **not use EDTA, heparan or citrate tubes or capillaries to collect the blood.** 4. Sample viability (if stored according to protocol in plastic bag with desiccant): 3 months.
Information Required with sample(s):	Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequen 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 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:	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
Reference ranges & units:	Referance range available upon request. Unit: nmol/mL/hr
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

PLEASE NOTE: Collection, courier and administration costs are included.
Protocol for each individual test is available on our website: www.pliem.co.za
Valid: 1 January 2022 - 31 December 2022