Centre for Human Metabolomics (CHM)

Test:	**Selective Lysosomal disorder screening - BLOOD SPOT SAMPLES (2x Guthrie cards)**
Test Code:	4268 x2
Tariff (including VAT):	R 1922.00 x 2 = R 3844.00
Description:	**Lysosomal storage disorders screen:** Fabry disease (α-galactosidase), Krabbe disease/globoid cell leukodystrophy / galactosylceramide lipidosis (galactocerebrosidase), Gaucher disease (ß-glucosidase), Niemann-Pick disease A / B (sphingomyelinase), MPS I / Hurler-, Hurler-Scheie-, Sheie syndrome (α-L-iduronidase), Pompe / Glycogen storage defect type II (α-glucosidase).
Turnaround time:	1. **30 work days** from receipt of sample at our laboratory (excluding public holidays and weekends). 2. Abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease. 3. To **verify** a **preliminary diagnosis** based on the analysis, **independent molecular genetic analyses are required** (also done on blood cards).
Transit stability / Sample viability:	**1. Do not expose specimen to heat or direct sunlight**. 2. Do not stack wet specimens. 3. Keep specimen dry. 4. **AFTER the blood card samples** are completely **dry**, ensure that it is **transported within 36 hours after collection** to PLIEM laboratory. 5. **If the samples are exposed to heat and not handled according to PLIEM laboratory's protocol, the samples will not be viable for the testing.**
Comments:	Specimens exposed to heat >25 ° C will not be viable for testing. Blood transfusion prior to collection may influence the analysis. Test is performed at accredited external laboratory in the USA.
Sample required:	**1. 2 x Blood collection cards (Guthrie cards / DBS)** by heal prick or fingerstick for a patient > 1 year of age. 2. Allow blood to **dry on the filter paper at ambient temperature** in a **horizontal position for 3 hours. Required: Whatman Protein Saver 903 Paper**.
Method:	Enzyme assay on tandem mass spectrometry
Reference ranges & units:	Gaucher: Beta-Glucosidase > or = 1.75 nmol/mL/hr Niemann-Pick A/B : Sphingomyelinase > or = 2.5 nmol/mL/hr Pompe disease: Alpha-Glucosidase > or = 3 nmol/mL/hr Fabry: Alpha-Galactosidase > or = 2.75 nmol/mL/hr MPS I: Alpha-L-Iduronidase > or = 2.0 nmol/mL/hr Krabbe: Galactocerebrosidase > or = 0.4 nmol/mL/hr
Consultant/Scientist:	Dr Marli Dercksen / Prof Chris Vorster
Telephone number:	018 299 2302 / 018 299 4196
E-mail address:	marli.dercksen@nwu.ac.za / chris.vorster@nwu.ac.za
Contact for results & other enquiries:	Ansie Mienie
Telephone number:	018 299 2312 / 018 299 1815 / 018 285 2652 (leave message)
Fax number:	182 992 316
E-mail address:	ansie.mienie@nwu.ac.za
Delivery address for samples:	Att: Dr M Dercksen, School for Biochemistry, PLIEM LAB POTCH, Building F3, Laboratory G20 (ground floor), 11 Hoffmann street, North West University, Potchefstroom

PLEASE NOTE: Collection, courier and administration costs are not included.
Protocol for each individual test is available on our website: www.pliem.co.za
Valid: 1 January 2018 - 31 December 2018