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Centre for Human Metabolomics (CHM)

Test:	**Selective Lysosomal disorder screening [PLSD] - BLOOD SPOT SAMPLES [DBS] (1 x Guthrie card)**
PLIEM Mnemonic:	**4268 - PPLSDg**
	Genzyme - PPLSDgPRO
NHRPL Tariff code	4268 (private - referred via path lab)
Tariff (including VAT):	R2 343.16 (referred via path lab)
	No charge (referred via Genzyme)
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. 10 - 15 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. Genetic testing Is required to confirm the diagnosis of a related disorder
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.
Sample required:	1. 1 x Blood collection card (Guthrie card / DBS) by heal prick (<1 year) of age OR fingerstick (> 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. **3. If blood is not applied directly onto the filter paper, do not use EDTA or citrate tubes or capillaries to collect the blood.**
Information Required with sample(s):	Absent clinical details may affect the interpretation of results and recommendations for further/additional testing (to assist with a differential diagnosis) cannot be made. 1. Clinical history of the patient. The referring clinician could complete and submit the clinical history on our website at https://pliem.co.za/test-request-form OR download the clinical history form from our website (same link) and email the completed form back to our laboratory at ansie.mienie@nwu.ac.za / pliem@nwu.ac.za. 2. Other significant medical reports for the patient (e.g. MRI brain, EEG, X-Ray reports, sonar reports, biopsy reports, genetic testing reports, etc). The referring clinician must please email these additional reports to ansie.mienie@nwu.ac.za. 3. Cumulative, routine pathology results of the patient (including archive results available) - this must be provided to our laboratory by the referring pathology laboratory. It could be e-mailed to pliem@nwu.ac.za OR send together with the sample(s) of the patient.
Method:	(Validated method Nov. 2019)
	Quantitative measurement of six lysosomal storage disorder (LSD) enzyme activities in DBS via liquid chromatography with 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
Contact for results & other enquiries:	Sample reception and resulting
Telephone number:	018 299 2312 / 018 285 2652 (leave message)
Fax number:	018 299 2316
E-mail address:	pliem@nwu.ac.za

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 2021 - 31 December 2021

Centre for Human Metabolomics (CHM)

Delivery address for samples:	Center for Human Metabolomics (CHM), Sample reception (PLIEM/NBS/CRS) Building F3, Room Number G19, 11 Hoffmann street North West University, Potchefstroom, 2531

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