

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 (β -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 heel 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 Galactocerebrosidase > or = 0.4 nmol/mL/hr Krabbe:
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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

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