

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 5/1/1965
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Leukocyte Lysosomal Enzyme Screen

ARUP test code 0099603

Leukocyte Lysosomal Enzyme Screen

See Note

SUBSTRATE/ ENZYME ACTIVITY: -----	DISEASE: -----	TISSUE: -----	RESULT: -----
4MU-b-D-galactoside b-galactosidase	GM1 gangliosidosis	L	72.2
4MU-b-D-mannoside b-mannosidase	b-Mannosidosis	L	100.0
4MU-a-L-fucoside a-L-fucosidase	Fucosidosis	L	40.3
4MU-a-D-mannoside a-mannosidase	a-Mannosidosis	L	188.0
4MU-b-D-glucuronide b-glucuronidase	MPS VII		
4MU-b-D-NACglCSO4 b-hexosaminidase A	Tay-Sachs and variants	L	227.8
Nitrocatechol sulfate Arylsulfatase A	Metachromatic leukodystrophy	L	84.3
Galactosylceramide Galactocerebrosidase	Krabbe disease	L	1.6
Sphingomyelin Sphingomyelinase	Niemann-Pick disease Types A and B		
4MU-b-D-glucoside Glucocerebrosidase	Gaucher disease		
4MU-a-L-iduronide a-L-iduronidase	MPS I		
Sialic acid content	Sialidosis, Sialuria, SASD		
4MU-a-NANA (fibro only) Sialidase	Sialidosis		
Nitrocatechol sulfate Arylsulfatase B	Maroteaux-Lamy syndrome		

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

4MU-a-D-galactoside
a-galactosidase Fabry disease

4MU-a-D-Nacglucosamine
a-glucosaminidase Sanfilippo Type B

4MU-b-D-Nacglucosamine
b-hexosaminidase
(Total and % A) Sandhoff and
Tay-Sachs

4MU-a-D-glucoside
a-glucosidase Pompe disease

4MU-palmitate
Acid lipase wolman disease/CESD

Mucopolidoses II and III (if indicated) ruled out by normal plasma enzymes:

Diagnosis: All values within normal limits

Signed: Paola Luzi, PhD

Abbreviations: 4MU=4methylumbelliferyl; NANA=N-Acetylneuraminic Acid; L=leukocytes; P=Plasma or Serum; F=fibroblasts; CVS=chorionic villi sample; AFC=amniotic fluid cells; CT=cultured trophoblasts; U=urine; NES=not enough sample; Units: L, F, CVS, AFC, CT=nmol/hour/mg protein; P=nmol/hour/ml; Sialic acid content=nmol/mg protein; Pompe disease=ratio of a-glucosidase activity with/without acarbose added.

These data were developed and validated by this laboratory. They have not been approved by the FDA and are to be used for clinical purposes.

Performed By: Jefferson Med College Lysosomal Lab
1020 Locust St
Rm 346
Philadelphia, PA 19107

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Leukocyte Lysosomal Enzyme Screen	23-214-100922	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at: