

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 3/7/2023
Gender: Male
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:
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4MU-b-D-galactoside b-galactosidase	GM1 gangliosidosis	L	61.6
4MU-b-D-mannoside b-mannosidase	b-Mannosidosis	L	45.0
4MU-a-L-fucoside a-L-fucosidase	Fucosidosis		
4MU-a-D-mannoside a-mannosidase	a-Mannosidosis		
4MU-b-D-glucuronide b-glucuronidase	MPS VII		
4MU-b-D-NACglcSO4 b-hexosaminidase A	Tay-Sachs and variants	L	129.9
Nitrocatechol sulfate Arylsulfatase A	Metachromatic leukodystrophy	L	32.0
Galactosylceramide Galactocerebrosidase	Krabbe disease	L	1.8
Sphingomyelin Sphingomyelinase	Niemann-Pick disease Types A and B	L	1.3
4MU-b-D-glucoside Glucocerebrosidase	Gaucher disease	L	2.4
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	L	328
4MU-a-D-glucoside a-glucosidase	Pompe disease	L	0.10
4MU-palmitate Acid lipase	wolman disease/CESD		

The sample received was slightly hemolyzed. All values are at the lower end of the unaffected range, except alpha-glucosidase activity. The ratio of alpha-glucosidase activity measured with and without acarbose is within the affected range for Pompe disease. We suggest to perform mutation analysis to confirm this diagnosis.

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

Diagnosis: Pompe disease-pending confirmation

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	23-191-401452	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: