

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 5/22/2014
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Pompe Disease (GAA), Enzyme Activity in Leukocytes

ARUP test code 2014463

Pompe GAA Activity Leukocytes Interp

See Note

In this sample, lysosomal acid alpha-glucosidase (GAA) activity was markedly reduced. The ratio between GAA activity and the total glucosidase activity, which includes interfering glucosidases, was also low (0.28; normal range: 0.3-0.67). These results are consistent with Pompe disease (OMIM#606800, Glycogen Storage Disease II). Consideration should be given to DNA testing to confirm this finding. Genetic evaluation is recommended.

This test cannot predict disease severity. Additionally, this test does NOT evaluate enzyme activities deficient in other lysosomal storage disorders.

Results reviewed and interpreted by [REDACTED]

INTERPRETIVE INFORMATION: Pompe Disease (GAA), Enzyme Activity in Leukocytes

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Pompe GAA Activity Leukocytes

0.8 nmol/h/mg L (Ref Interval: 5.5-25.0)

Units of measure for acid alpha-glucosidase (GAA) activity (Pompe Disease) are: Nanomoles of substrate hydrolyzed per hour per mg of protein.

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

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Pompe GAA Activity Leukocytes Interp	24-144-116286	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Pompe GAA Activity Leukocytes	24-144-116286	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:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 24-144-116286
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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