

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** Unknown  
**Gender:** Unknown  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Hexosaminidase A Percent and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A Percent and Total Hexosaminidase in Leukocytes**

ARUP test code 2008129

**Hexosaminidase Total** 791.0 (Ref Interval: 600.0-1050.0)  
INTERPRETIVE INFORMATION: Hexosaminidase A Pct and Total in P/S  
Total Hexosaminidase reported in nmols hydrolyzed/hr/mL  
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.

**Hexosaminidase A Percent** 53.4 % L (Ref Interval: 55.0-76.0)

**Hexosaminidase Interpretation** See Note  
In this sample, serum total hexosaminidase activity was normal with hexosaminidase A activity, expressed as percentage of total hexosaminidase, outside the normal range (normal range: 55 - 76%) but higher than observed in Tay-Sachs disease (OMIM 272800) carriers (TSD carrier range < 51%). Carrier status for Tay-Sachs disease is possible in this patient. To exclude this possibility, testing in leukocytes was performed. Leukocyte total hexosaminidase activity was within normal ranges. Hexosaminidase A activity in leukocytes was at the upper limit of the carrier range for Tay-Sachs disease. Based on these results, this individual is NOT affected with Tay-Sachs disease (OMIM 272800). Carrier status for Tay-Sachs disease cannot be excluded. Consideration should be given to mutational analysis to confirm carrier status and exclude pseudo-deficiency (Tay-Sachs Disease (HEXA) Sequencing and 7.6kb Deletion, ARUP test code 2009298). Pseudo-deficiency has been shown for this enzyme, due to specific mutations affecting the exogenous substrate used by this assay, but not endogenous substrates. Milder variant forms of Tay-Sachs disease, such as the B1 variant or the activator protein deficiency (OMIM 272750), may not be identified by this assay.

**Hexosaminidase A Percent and Total in Leukocytes (Internal Only)**

ARUP test code 2008470

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

Hexosaminidase Total, Leukocytes

1786.0

INTERPRETIVE INFORMATION: Hexosaminidase A Pct and Total in Leukocytes

Total Hexosaminidase in Leukocytes is reported in nmols hydrolyzed/hr/mg of protein.

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Hexosaminidase A Percent in Leukocytes

**59.3 % L**

(Ref Interval:  $\geq 63.0$ )

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Hexosaminidase Total	21-120-122777	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase A Percent	21-120-122777	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase Total, Leukocytes	21-120-122777	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase A Percent in Leukocytes	21-120-122777	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase Interpretation	21-120-122777	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