

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, Plasma or Serum

ARUP test code 2008121

Hexosaminidase Total	2986.0	H	(Ref Interval: 600.0-1050.0)
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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	30.4 %	L	(Ref Interval: 55.0-76.0)
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Hexosaminidase Plasma/Serum Interp	See Note		
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In this sample, hexosaminidase A activity, expressed as percentage of total hexosaminidase, was reduced and within the carrier range for Tay-Sachs disease (OMIM 272800). Total hexosaminidase activity was markedly elevated, which likely reflects elevated levels of alternative, heat-resistant forms of hexosaminidase. Several conditions, such as pregnancy increase total hexosaminidase activity in serum/plasma, but not in leukocytes. Based on these results, carrier status for Tay-Sachs disease is possible in this patient; however, the elevated levels of hexosaminidase can cause false positive results. Would repeat testing in leukocytes to exclude this possibility (Hexosaminidase A and Total hexosaminidase in Leukocytes, ARUP test code 2008125). 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.

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

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Hexosaminidase Total	21-120-122580	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase A Percent	21-120-122580	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hexosaminidase Plasma/Serum Interp	21-120-122580	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
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 21-120-122580
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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