

Client: Example Client ABC123

123 Test Drive

Salt Lake City, UT 84108

UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example** 

DOB 4/19/2022 Female Sex:

**Patient Identifiers:** 01234567890ABCD, 012345

**Visit Number (FIN):** 01234567890ABCD **Collection Date:** 01/01/2017 12:34

## Alpha-Iduronidase Enzyme Activity in Leukocytes

ARUP test code 2011415

## Alpha-Iduronidase, Leukocytes, Interp

## See Note

In this sample, alpha-L-iduronidase activity was markedly reduced, and suggestive of a diagnosis of Mucopolysaccharidosis type I (Hurler/Hurler-Scheie/Scheie), although alpha-L-iduronidase activity is typically undetectable in these patients. These levels have been described in carriers of pseudodeficiency alleles. Pseudodeficiency has been shown for this enzyme due to specific variants affecting the exogenous substrate used by this assay, but not endogenous substrates. Consideration should be given to molecular testing of the IDUA gene to exclude MPSI or pseudodeficiency. Genetic evaluation is recommended. This test cannot predict disease severity. Additionally, this test does NOT evaluate enzyme activities deficient in other mucopolysaccharidoses.

Units are Nanomoles substrate hydrolyzed per hour per milligram of protein (nmoles hydrolyzed/hr/mg protein)

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.

## Alpha-Iduronidase Activity, Leukocytes

1.1

(Ref Interval: 12.0-65.0)

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Alpha-Iduronidase, Leukocytes, Interp	22-123-400152	5/2/2022 4:13:00 PM	5/4/2022 6:51:45 AM	5/9/2022 8:25:00 AM
Alpha-Iduronidase Activity, Leukocytes	22-123-400152	5/2/2022 4:13:00 PM	5/4/2022 6:51:45 AM	5/9/2022 8:25:00 AM

L

END OF CHART

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