

500 Chipeta Way, Salt Lake City, Utah 84108-1221

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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Patient Age/Sex: 10 years Male

Specimen Collected: 19-Jun-24 16:54

Alpha-Iduronidase, Leukocytes Procedure	Received: 19-Jun-24 16:54 Result	Report/Verified: 19-Jun-24 17:00 Units	Reference Interval
Alpha-Iduronidase, Leukocytes, Interp	See Note ^{f1 i1}		
Alpha-Iduronidase Activity, Leukocytes	1.1 ^{L i2}	nmol/h/mg	[12.0-65.0]

Result Footnote

f1: Alpha-Iduronidase, Leukocytes, Interp
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.

Results reviewed and interpreted by [REDACTED]

Test Information

i1: Alpha-Iduronidase, Leukocytes, Interp
INTERPRETIVE INFORMATION: Alpha-Iduronidase, 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.

i2: Alpha-Iduronidase Activity, Leukocytes
Units of measure for alpha-L-iduronidase activity (Mucopolysaccharidosis type I) are: Nanomoles of substrate hydrolyzed per hour per mg of protein.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, i-Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

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