Alpha-Iduronidase Enzyme Activity in Leukocytes

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Disease Overview

Mucopolysaccharidosis type I (MPS I) is a progressive disorder that ranges in severity and can affect numerous systems throughout the body. Subtypes include attenuated MPS I (previously known as Hurler-Scheie and Scheie syndromes) and severe MPS I (formerly Hurler syndrome). Symptoms vary widely but can include cardiomyopathy, “coarse” facial features (eg, thickening of the earlobes, lips, outside of nostrils, and tongue), intellectual disability, organomegaly, and progressive skeletal dysplasia.[1]

Featured ARUP Testing

Alpha-Iduronidase Enzyme Activity in Leukocytes 2011415

Method: Quantitative Fluorometry

Recommended test to exclude MPS I following an abnormal screen or to confirm MPS I in patients with a consistent clinical phenotype and/or a positive family history

For additional MPS testing, refer to the Laboratory Test Directory.

Genetics

Gene

IDUA

Inheritance

Autosomal recessive

Incidence

MPS I: Approximately 1/70,000

- Attenuated MPS I: 1/500,000[1]
- Severe MPS I: 1/100,000[1]

Genotype/Phenotype Correlation

MPS I Subtype	Characteristics
Attenuated MPS I	Variants are often milder (eg, single base-pair substitutions) • Some residual enzyme function is retained
Severe MPS I	Associated with more severe, nonsense variants • Profound alpha-iduronidase deficiency

Test Interpretation

Results

An extremely low or undetectable level of alpha-iduronidase enzyme activity in leukocytes is consistent with MPS I.

Limitations

Testing for alpha-iduronidase enzyme activity:

- Cannot differentiate between attenuated MPS I (ie, Hurler-Scheie and Scheie syndromes) and severe MPS I (Hurler syndrome)
  - Categorization depends on clinical and/or molecular genetic findings
- Cannot predict carrier status for MPS I
- Does not evaluate enzyme deficiencies in other MPS types

Additionally, pseudodeficiency has been demonstrated for this enzyme due to specific gene variants that affect the exogenous substrate but not endogenous substrates. Individuals with pseudodeficiency are not affected with MPS I.

References

1. Clarke LA. Mucopolysaccharidosis type I. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews*, University of Washington, Seattle. Last update Feb 2021; accessed Jan 2022.

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