

Notification Date: September 18, 2023 Effective Date: October 17, 2023

Iduronate-2-Sulfatase, Leukocytes

Test ID: 12SWB

Useful for:

Supporting the biochemical diagnosis of mucopolysaccharidosis II (MPS II; Hunter syndrome) in whole blood specimens

This test is **not useful for** carrier detection for MPS II.

Genetics Information:

This test provides diagnostic testing for individuals with positive newborn screen results or clinical signs and symptoms suspicious for mucopolysaccharidosis type II (MPS II, Hunter syndrome). If an enzyme deficiency is

Methods:

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

Reference Values:

>2.20 nmol/hour/mg protein.

An interpretive report will be provided.

Specimen Requirements:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot.**

Minimum Volume: 5 mL

Specimen Stability Information:

Specimen Type	Temperature	Time
Whole Blood ACD	Refrigerated (preferred)	6 days
	Ambient	6 days

Cautions:

Individuals with pseudodeficiency alleles can show reduced enzyme activity.

Carrier status (heterozygosity) for these conditions cannot be reliably detected.

Enzyme levels may be normal in individuals receiving enzyme replacement therapy or who have undergone hematopoietic stem cell transplant.

Iduronate-2-sulfatase can also be deficient in individuals with multiple sulfatase deficiency.

CPT Code:

82657

Day(s) Performed:

Report Available:

Preanalytical processing: Monday through Saturday 8

8 to 15 days

Testing performed: Tuesday

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.