

## Mucopolysaccharidosis IV Enzyme Panel, Leukocytes

**Test ID:** MPS4W

### Useful for:

Supporting the biochemical diagnosis of mucopolysaccharidosis type IVA and IVB in whole blood specimens

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

### Genetics Information:

This test provides diagnostic testing for individuals with clinical signs and symptoms suspicious for mucopolysaccharidosis type IVA or IVB. If an enzyme deficiency is detected by this test, additional biochemical or molecular testing is required to confirm a diagnosis.

### Methods:

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

### Reference Values:

N-acetylgalactosamine-6-sulfatase: >1.60 nmol/hour/mg protein

Beta-galactosidase: >0.28 nmol/hour/mg protein

An interpretive report will be provided.

### Specimen Requirements:

<b>Preferred:</b>	Yellow top (ACD solution B)
<b>Acceptable:</b>	Yellow top (ACD solution A) or lavender top (EDTA)
<b>Specimen Volume:</b>	6 mL
<b>Collection Instructions:</b>	Send whole blood specimen in original tube. <b>Do not aliquot.</b>
<b>Minimum Volume:</b>	5 mL

**Specimen Stability Information:**

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

**Cautions:**

Beta-galactosidase is also reduced in patients with galactosialidosis. Those patients will also demonstrate deficient activity of neuraminidase which is not evaluated on this panel. If there is clinical suspicion of galactosialidosis, please order follow up testing to include test OLIGU / Oligosaccharide Screen, Random, Urine

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.

**CPT Code:**

82657

**Day(s) Performed:**

Preanalytical processing: Monday through Saturday  
Testing performed: Tuesday

**Report Available:**

8 to 15 days

**Questions**

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