

## Endogenous Mucopolysaccharidosis Type II (I2S [Iduronate-2-Sulfatase]) Biomarker Reflex, Blood Spot

**Test ID:** MPS2R

### Useful for:

Second-tier testing of newborns with an abnormal primary screening result for mucopolysaccharidosis type II (MPS II, decreased iduronate-2-sulfatase) where quantitation of the glycosaminoglycans dermatan and heparan sulfate is desired in the presence of elevated endogenous MPS II biomarkers

Follow-up testing for evaluation of an abnormal newborn screening result for MPS II.

This test is **not a useful** monitoring test for individuals with MPS II.

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

### Genetics Information:

This test is a second-tier assay for newborns and infants who have abnormal newborn screening results for mucopolysaccharidosis type II (MPS II) with reduced iduronate-2-sulfatase activity.

### Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
MPSBS	Mucopolysaccharidosis, BS	Yes	No

### Method:

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

### Reference Values:

An interpretive report will be provided

## Specimen Requirements:

- Supplies:** Card-Blood Spot Collection (Filter Paper) (T493)
- Preferred:** Blood Spot Collection Card
- Acceptable:** Whatman Protein Saver 903 paper, PerkinElmer 226 filter paper, Munktell filter paper, local newborn screening card, postmortem screening card, or blood collected in tubes containing ACD or EDTA and dried on filter paper.
- Specimen Volume:** 2 Blood spots
- Collection Instructions:**
1. An alternative blood collection option for patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
  2. Completely fill at least 2 circles on filter paper card (approximately 100 microliters blood per circle).
  3. Let blood dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
  4. Do not expose specimen to heat or direct sunlight.
  5. Do not stack wet specimens.
  6. Keep specimen dry.
- Minimum Volume:** 1 Blood spot

## Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Whole Blood	Refrigerated (preferred)	30 days	FILTER PAPER
	Ambient	13 days	FILTER PAPER
	Frozen	90 days	FILTER PAPER

## Shipping Instructions:

Specimens stored at ambient temperatures for more than 13 days after collection may result in false-positive results in carrier and other unaffected individuals.

## Cautions:

Individuals with multiple sulfatase deficiency may also have abnormal levels of mucopolysaccharidosis type II specific biomarker and heparan and dermatan sulfate.

## CPT Code:

82542

**Day(s) Performed:** Mondays and Thursdays

**Report Available:** 3 to 6 days

## Questions

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