

Endogenous Mucopolysaccharidosis Type I (IDUA [Alpha-L-Iduronidase]) Biomarker Reflex, Blood Spot

Test ID: MPS1R

Useful for:

Second-tier testing of newborns with an abnormal primary screening result for mucopolysaccharidosis type I (MPS I, decreased alpha-L-iduronidase activity) where quantitation of the glycosaminoglycans dermatan and heparan sulfate is desired in the presence of elevated endogenous MPS I biomarkers

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

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

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

Genetic Information:

This test is a second-tier assay for newborns and infants who have abnormal newborn screening results for mucopolysaccharidosis type I (MPS I) with reduced alpha-L-iduronidase 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:

No significant cautionary statements

CPT Code:

82542

Day(s) Performed: Monday, Thursday

Report Available: 3 to 6 days

Questions

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