A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” or “critical” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

**Disorder Indicated:** MPS-1 is a Lysosomal Storage Disorder (LSD) where the body lacks sufficient activity of the **lysosomal enzyme alpha-L-iduronidase (IDUA)**, caused by mutations in the **IDUA gene**, and results in accumulation of glycosaminoglycan (GAG) in the lysosomes resulting in swelling, cell damage, and progressive organ dysfunction.

**1st Tier Testing**

Screens meeting criteria (critical, multiple abnormal screens) will move to 2nd Tier testing

**Analyte Measured:** Enzyme activity: lysosomal enzyme alpha-L-iduronidase (IDUA)

**Normal Test Results:** See report for additional information

**Abnormal Test Results:** See report for additional information

**Critical Test Results:** See report for additional information

**Incidence:** 1 in every 100,000 newborns.

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**Signs and Symptoms may include:**

- Umbilical or inguinal hernia may be present
- Valvular heart disease, Cardiac failure
- Frequent respiratory infections
- Progressive skeletal dysplasia and other abnormalities
- Coarse facial features, spinal deformity
- Hearing loss
- Significant, progressive intellectual or learning difficulties
- Death from cardiorespiratory complication within 1st decade if untreated

**MPS-1 Attenuated typically has less severe symptoms of above and progresses more slowly**

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**Next Steps may include:**

Discuss the next steps of evaluation and possible treatment with the regional Geneticist

Provide parental education (see accompanying sheet)

Clinical Assessment

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**Treatment (if indicated):** Enzyme Replacement Therapy and Hematopoietic stem cell transplantation (HSCT)

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**Additional Resources**

VDH Newborn Screening [http://vdhlivewell.com/newbornscreening](http://vdhlivewell.com/newbornscreening)

Baby’s First Test [www.babysfirsttest.org](http://www.babysfirsttest.org)

American College of Medical Genetics (ACMG) ACT Sheets [www.ACMG.net](http://www.ACMG.net)


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**Educational content adapted from** [www.babysfirsttest.org](http://www.babysfirsttest.org)