Mucopolysaccharidosis Type-I (MPS-1)

**Parent Fact Sheet**

A newborn screening test is a *screen* and not diagnostic testing. An “abnormal” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is *very important* to determine if your baby has the disorder indicated.

**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.

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<th>Incidence</th>
<th>1 in every 100,000 newborns.</th>
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<td>Signs and Symptoms</td>
<td>When a child has MPS-1, you may see symptoms including:</td>
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| *Please note: these findings may not be present in young infants or in milder forms of the disease* | - 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*

| Next Steps *may* include: | Follow up with your child’s pediatric provider  
Clinical assessment  
Laboratory Testing |

**Treatment (if indicated)** | Follow up with your child’s pediatric provider and Genetic Specialist |

**Additional Resources** | VDH Newborn Screening [http://vdhlivewell.com/newbornscreening](http://vdhlivewell.com/newbornscreening)  
Baby’s First Test [www.babysfirsttest.org](http://www.babysfirsttest.org)  

*Educational content adapted from www.babysfirsttest.org*