

## Mucopolysaccharidosis Type-I (MPS-1)

### Parent Fact Sheet

<p>A newborn screening test is a <b>screen</b> 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 <b>very important</b> to determine if your baby has the disorder indicated.</p>	
<p><b>Disorder Indicated:</b> MPS-1 is a Lysosomal Storage Disorder (LSD) where the body lacks sufficient activity of the <i>lysosomal enzyme alpha-L-iduronidase</i> (IDUA), caused by mutations in the <i>IDUA</i> gene, and results in accumulation of glycosaminoglycan (GAG) in the lysosomes resulting in swelling, cell damage, and progressive organ dysfunction.</p>	
<p>Incidence</p>	<p>1 in every 100,000 newborns.</p>
<p>Signs and Symptoms</p> <p><i>Please note: these findings may not be present in young infants or in milder forms of the disease</i></p>	<p>When a child has MPS-1, you may see symptoms including:</p> <ul style="list-style-type: none"> <li>• Umbilical or inguinal hernia may be present</li> <li>• Valvular heart disease, Cardiac failure</li> <li>• Frequent respiratory infections</li> <li>• Progressive skeletal dysplasia and other abnormalities</li> <li>• Coarse facial features, spinal deformity</li> <li>• Hearing loss</li> <li>• Significant, progressive intellectual or learning difficulties</li> <li>• Death from cardiorespiratory complication within 1st decade if untreated</li> </ul> <p><i>MPS-1 Attenuated typically has less severe symptoms of above and progresses more slowly</i></p>
<p>Next Steps <b>may</b> include:</p>	<p><b>Follow up with your child’s pediatric provider</b></p> <p>Clinical assessment Laboratory Testing</p>
<p>Treatment <b>(if indicated)</b></p>	<p>Follow up with your child’s pediatric provider and Genetic Specialist</p>
<p>Additional Resources</p>	<p>VDH Newborn Screening <a href="http://vdhlivewell.com/newbornscreening">http://vdhlivewell.com/newbornscreening</a>          Baby’s First Test <a href="http://www.babysfirsttest.org">www.babysfirsttest.org</a>          Genetics Home Reference <a href="https://qhr.nlm.nih.gov/">https://qhr.nlm.nih.gov/</a>          National MPS Society <a href="http://www.mpssociety.org/">http://www.mpssociety.org/</a></p>

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

