

Maple Syrup Urine Disease (MSUD)

Parent Fact Sheet

<p>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.</p>	
<p>Disorder Indicated: Maple Syrup Urine Disease (MSUD) is a condition in which the body cannot break down certain parts of protein, including certain amino acids due to an enzyme deficiency. This results in an accumulation of the amino acids leucine, isoleucine and valine. If left untreated, MSUD could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with MSUD can have healthy growth and development.</p>	
<p>Incidence</p>	<p>1 in every 185,000 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 Maple Syrup Urine Disease, you can may see symptoms including:</p> <ul style="list-style-type: none"> • Urine that smells sweet like maple syrup • Poor appetite/ Weight loss/ Vomiting • Trouble sucking during feeding • High pitched cry • Sleeping longer or more often/Tiredness • Irritability • Developmental delays <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
<p>Next Steps may include:</p>	<p>Follow up with your child’s pediatric provider</p> <p>Clinical assessment Laboratory Testing</p>
<p>Treatment (if indicated)</p>	<p>Restricted diet (follow up with your child’s pediatric provider)</p>
<p>Additional Resources</p>	<p>VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org Genetics Home Reference https://ghr.nlm.nih.gov/ MSUD Family Support Group http://www.msud-support.org/</p>

Educational content adapted from www.babysfirsttest.org

